PSYCHOLOGICAL EFFECTS OF NEONATAL SCREENING

A study of parents of children at high risk for serious illness in adulthood due to alpha$_1$antitrypsin deficiency

by

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Title and subtitle PSYCHOLOGICAL EFFECTS OF NEONATAL SCREENING

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Abstract

Nation-wide neonatal screening for α_1-antitrypsin deficiency (ATD), which leads to chronic pulmonary disease in adulthood, was conducted in Sweden in 1972-74, but not continued thereafter due to observations of negative psychological effects on the parents and parent-child relationship. A systematic study of such effects was carried out when the children were 5-7 years old. The families of 61 clinically healthy children with ATD were studied using a number of methods, including parental interviews and observation of mother-child interaction in the home, use of population register and medical record data, etc.

The parents were informed of the child's ATD before it was 6 months old, and a majority of the parents reported having been shocked and frightened at the identification of the ATD; many of the mothers still evidenced such feelings toward the ATD at follow-up. Comparison with control families with normal children showed the mothers (but not the fathers) in the ATD-group to report significantly poorer mental and physical health at follow-up, which appeared to be a consequence of stress resulting from the identification. No long-term effects were found on the ATD-families' reproduction rates, marital status, social class level, parental attitudes towards themselves as parents, toward their current life situation or toward having more children.

The total results indicated that, as viewed from a psychological perspective, screening for ATD should not be reinstated in the same form as that in 1972-74.

Key words

Neonatal screening, alpha$_1$-antitrypsin deficiency, prevention, risk group, cost-benefit analysis, parents, emotional reactions, emotional adjustment, psychological effects

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INTRODUCTION

Primary prevention and screening

Progress in medical science during the last decades has greatly increased the possibilities for primary prevention of illness. Today many diseases can be identified at a presymptomatic stage, and some of these are also potentially preventable through early intervention. Investigative methods are increasingly available for early diagnosis of diseases or for identification of individuals at high risk for disease. These methods have provided the basis for screening, i.e. the use of a test or examination technique to identify those individuals in a population who probably have (or will develop) a given illness and those who probably are healthy (Whitby 1974). The term, mass-screening, is used if the method is applied to all individuals in a population.

A number of congenital diseases (usually inherited metabolic diseases) and predispositions for illness can at present be identified through screening in the neonatal period (Larsson 1980), and the number of these diseases is rapidly increasing (Boström & Ljungstedt 1980, Kaback 1978). Guidelines for mass health screening have been developed and published by WHO (Wilson & Jungner 1968) and recommendations for screening for congenital diseases have subsequently been developed by several groups (Raine 1977, Scriver 1976, Scriver et al 1977, Taussig 1983, WHO 1971).

These recommendations can be summarized briefly in the following way: A method with high sensitivity and specificity for (presymptomatic) identification of the disease should exist. The use of treatment or preventive measures following the early identification should lead to improved health in the population, as compared with that attained through traditional diagnosis and treatment procedures. The method should be easy and inexpensive to use and not harmful to the individual. The "benefits" of the screening for the disease in question (i.e. evidence of improved health, increased survival, lower treatment costs etc) should exceed the "costs" (e.g the costs for organization and methodology, possible unnecessarily negative effects on the individuals in the population) in order for the screening to be defensible, as indicated in the total cost-benefit analysis. An organization should be established to administer both the screening itself and the follow-up and treatment of identified cases in a competent manner.

Increased experience with screening has made it more and more apparent

that, in addition to screening's somatic and economic aspects, its ethical, psychological and psychosocial aspects also must be included in the total cost-benefit analysis (Kaback 1978, Lappé et al 1972, Larsson 1980, Raine 1977, Taussig 1983). Factors such as negative (and positive) psychological consequences for the families of identifying presymptomatic illness in children and effects of falsely positive identifications should enter into the cost-benefit analysis. This analysis should as far as possible be based on the "real" costs and benefits (as determined through careful empirical study), rather than on the theoretical or potential costs and benefits.

Alpha$_1$antitrypsin deficiency (ATD) - a predisposition for illness

Severe deficiency of $\alpha_1$antitrypsin (PiZ type, with about 15 % of the normal level of $\alpha_1$antitrypsin in the blood) has an incidence of 1/1500 in Sweden. Alpha$_1$antitrypsin is a blood protein that inhibits and inactivates the proteolytic enzymes normally existing in the individual's granulocytes, especially elastase in the lungs. ATD is inherited in an autosomal co-dominant manner. Individuals with severe ATD have a somewhat increased risk for neonatal liver disease and a greatly increased risk for chronic obstructive lung disease in adulthood (Eriksson et al 1980, Sveger 1981). Onset of the lung disease can be considerably postponed if the individual with ATD avoids exposure to concentrated air pollution, especially tobacco smoke. Smokers with severe ATD (PiZ) have an earlier symptom onset (median age 35-40 years) and reduced survival (median age at death about 50 years), compared with that for non-smokers with ATD (median age at onset about 50 years, at death about 65 years) (Janus et al 1985, Larsson 1978). While a clinically practicable treatment for ATD does not exist at present, preventive measures such as the avoidance of polluted air and abstaining from smoking represent considerable potential gains for the future health of individuals with ATD.

ATD exists during intrauterine life and at birth but most newborns with ATD are asymptomatic and lack externally observable signs of the deficiency, except for the approximately 10 % who have neonatal liver disease (Sveger 1976). ATD can be identified in the neonatal period by means of an easily applied and inexpensive test with very high sensitivity and specificity for ATD of PiZ type, using the same blood sample as for PKU (Laurell 1972).

Neonatal screening for ATD 1972-1974

During the period, November 1972 - September 1974, a pilot screening for ATD was performed on all newborns in Sweden (n = 200,000). In total, 171 children with ATD were identified, and their types and levels of ATD were confirmed through new blood tests at about three months of age; 120 children had the homozygous PiZ type with about 15% of the normal level, while the majority of the remaining cases had the heterozygous PiSZ type with about 40 % of the normal level (Sveger 1976).

In association with the new blood test at about three months after birth, the parents were informed about the child's ATD and about the relevance of smoking for the development of lung disease in the future. Parents who smoked were advised to stop. Almost all children (and their parents) participated thereafter in a follow-up study, with planned check-ups by a pediatrician at 1/2, 1, 2 and 4 years of age, at which the children were examined physically and laboratory tests performed (Sveger 1976, Sveger & Thelin 1981).

In association with the follow-up of the children, a number of pediatricians observed that the identification of the ATD had had notably negative psychological effects on some of the parents. In some cases, information about the child's ATD appeared to have evoked strong anxiety and negatively influenced the parent-child relationship. Some pediatricians thought that the neonatal period represented an especially sensitive time for parents and was therefore not an optimal time for informing them that something is wrong with their child.

ATD-screening was not continued after the pilot study terminated in 1974. The Swedish Pediatrics Association (Svenska Paediatriska Föreningen) and the Swedish Medical Board (Socialstyrelsen) recommended that the psychological consequences for the family of this pilot neonatal ATD-screening be assessed before making a decision regarding future screening for ATD.

The present study's purpose and relevance

An investigation of the ATD-screening's psychological and psychosocial consequences for the families of children with ATD was begun when the children were 5-7 years old.

The primary goal for the study in total was to investigate whether the "ATD-matter" (the child's ATD, the screening, the information to the parents, ATD-related contacts with medical services, blood tests etc.) had a/ had a negative effect on the parents' attitudes and feelings toward

themselves as parents and toward the child, and b/ negatively influenced the parent-child relationship and selected characteristics of the child. The study also intended to describe systematically c/ what had happened in association with the medical system's contacts with the parents, d/ the parents' reactions in association with the identification and follow-up of the child's ATD, e/ the parents' current feelings, attitudes and knowledge associated with the child's ATD, and f/ the parents' recommendations regarding future screening for ATD.

The study's results should, if possible, contribute to increased and empirically-based knowledge relevant first for the decision about whether or not to reinstate the ATD-screening in the previous form, and second for how such screening and follow-up of children with ATD and their families could be improved from a psychological perspective (if it were reinstated). The latter goal can best be attained by also studying factors that influenced the individual parent's reactions and adjustment to the child's ATD.

The study's results are relevant for screening not only for ATD per se but also for other disorders. The possibilities for identifying other groups that are at high genetic risk for disease have increased (Boström & Ljungstedt 1980, Kaback 1978) and knowledge about the psychological consequences of screening has come to be viewed as increasingly important (Boström & Ljungstedt 1980, Larsson 1980, McNeil & Thelin 1980, Raine 1977, Schild 1979, Scriver 1976, Taussig 1983). Furthermore, few systematic empirical studies of psychological effects of screening have thus far been done, and the current study's design, hypotheses and methods can possibly be useful in studying similar questions regarding other diseases and risk conditions.

Goal of the dissertation

The purpose of this dissertation is to provide answers to those questions that concern the psychological and psychosocial effects of neonatal ATD-screening on the parents. The parents' reports of their experiences of the contacts with the medical system concerning their children's ATD and their recommendations for renewed screening are also presented.

The decision to concentrate on the effects on the parent (rather than on the parent-child relationship or the child) was based on the fact that the initiative for the total study resulted from the pediatricians' original observations of negative effects on the parents. Further, with the

ATD-identification in the neonatal period, any possible effects on the parent-child relationship or on the child would have to have been mediated through the effect on the parents.

The following questions are addressed in this dissertation (the Roman numbers in parentheses refer to the specific articles presented in this dissertation):

Question 1. How can the psychological and psychosocial effects on the parents of neonatal identification of ATD be studied, 5-7 years later? (I)

Question 2. How were the parents first contacted about the child's ATD and how did they react to this contact? (II)

Question 3. How did the parents experience the subsequent contacts with the medical system concerning the child's ATD? (III)

Question 4. What current feelings and degree of emotional adjustment toward the child's ATD did the parents show 5-7 years after its identification? (IV)

Question 5. What recommendations did the parents have concerning a new screening for ATD? (III)

Question 6. Had the ATD-matter influenced the parents' demographic characteristics (reproduction, marital status and social class level)? (V)

Question 7. Had the ATD-matter negatively influenced the parents' attitudes a/ toward themselves as parents, b/ toward having more children (VI) and c/ toward their current life situation; and had it influenced their experienced mental and physical health? (VII)

Psychological effects of neonatal screening on the parents

At the time of planning this study, no other systematic studies of the psychological effects on families with children who were screened in the neonatal period for high risk for future illness were known to exist. A pilot mass-screening for ATD has been conducted in the neonatal period in the USA (O'Brien et al 1978), but the possible psychological consequences of this screening were not studied. Continuation of that ATD-screening was not recommended, primarily because of the low frequency of ATD in the USA and the absence of effective treatment for ATD.

The possibility that neonatal screening can lead to permanent, harmful psychological effects on the parents and child has been suggested (Solnit 1976). Lozoff et al (1977) have noted that the neonatal period can be an

especially sensitive time for intervening medically and informing the parents that something is wrong with their child.

Rothenberg and Sills (1968) observed that falsely positive PKU-screening results for newborns created strong anxiety in a considerable number of the parents. He emphasized the importance of offering these parents emotional support.

Bodegård et al (1983) have studied the psychological effects on the parents of falsely positive screening results for congenital hypothyroidism in neonates. Many of these parents had reacted with worry and anxiety when this disease was first suspected in the child, and about 20 % of the families were judged 6 months-1 year later to show signs of remaining psychological problems. Other children with falsely positive results for congenital metabolic diseases have been studied concerning parents' reactions both before and after a repeated test for confirmation of the existence of a disease (Sorenson et al 1984). The second test, which indicated normality, reduced the parents' anxiety and depressive feelings that had been elicited by the first, falsely positive result; however, 36 % of the parents still felt concern about the child's health even after the child had been found to be healthy.

ATD as a phenomenon - a comparison with illness

ATD per se is not an illness but rather a predisposition for illness. With the exception of those cases with neonatal liver disease (who were excluded from the current study), children with ATD are free from symptoms and lack external visible signs of the deviation. Nevertheless, from a psychological point of view, ATD in children has many similarities to disease: ATD represents a/ a measurable somatic deviation, which b/ was identified by the medical system and c/ led to physical examinations by a physician, blood tests and repeated check-ups of the child. The ATD might imply d/ a "weakness" that leads to a number of "doctor's orders" and required actions by the child and family (avoid air pollution, abstain from smoking).

Psychological effects of children's illness on the parents

As a phenomenon, ATD in children can be likened to illness in children. Much of the extensive literature on psychological effects on the parent of disease (chronic diseases, malformations etc) in children is relevant for constructing hypotheses about the long- and short-term psyc-

hological effects of the ATD-matter on the parents. Special interest has been given to those parts of the literature that can be of value to this study.

At the moment the parent becomes aware of his/her child's illness or deviation, a psychological process begins within the parent; this process is characterized by the parent's attempt to handle and adapt emotionally and practically to the new situation that the child's illness or deviation has created (Cullberg 1975, Janis 1974, Kaplan et al 1977, Myers et al 1977). The topics in the literature that have been given special interest represent both those that describe the parent's reactions and feelings resulting from becoming aware of the child's illness and those that concern observable manifestations (in feelings, attitudes, actions, health) of the parent's adaptive process that the child's illness has initiated.

Parents of children with serious or chronic illness show in general a strong psychological reaction when they become aware of the child's condition and realize its implications. These psychological reactions have often been viewed as representing a traumatic psychic crisis for the parent (Bocian & Kaback 1978, Caplan 1964, Drotar et al 1975, Laurell-Borulf 1982, Michaels & Schucman 1962, Rosen 1955). An extensive review of the literature on the topic (McNeil & Thelin 1980) indicated that parents' initial specific reactions can be quite varied. The most frequently described reactions were shock, confusion, guilt, depression, sadness, disappointment, anger, shame, denial, despair, and ambivalence toward and rejection of the child (Bocian & Kaback 1978, Carr & Oppé 1971, Chodoff et al 1964, Cummings et al 1969, Dodge et al 1978, Drotar et al 1975, Gilder et al 1978, Golden & Davis 1977, Graliker et al 1959, Mandelbaum & Wheeler 1960, Mattsson & Agle 1972, Michaels & Schucman 1962, Poznanski 1973, Roos 1963, Schild 1979, Solnit & Stark 1961, Wolfensberger 1967). The intensity and duration of the parent's reaction varies from case to case, depending both on the nature of the illness and on the parent's social situation and personal capacity to adapt psychologically to the new conditions that the child's illness represents.

In general, parents expect and fantasize about getting a well-developed, healthy child (Moos & Tsu 1977, Solnit & Stark 1961). When parents become aware that the child has an illness or deviation, and especially when this happens in the neonatal period, the parents often react not only to the illness and its possible consequences but also to their symbolic loss of the well-developed, healthy child they had fantasized about and

awaited. According to Solnit and Stark (1961), the parent must first mourn the loss of the "imaginary" healthy child before the conditions can exist for accepting and adapating to the "real", sick child. If this mourning process does not take place or is incomplete, it can lead to consequences for the parent such as long-lasting mental symptoms like anxiety, depression and feelings of guilt (Jackson 1977) and difficulty in relating to the child (Solnit & Stark 1961). Furthermore, the child's illness per se can represent a burden for the parent and contribute to the parent's mental problems (Michaels & Schucman 1962).

For many parents, a well-developed, healthy child symbolizes the parent's capability and sufficiency, and a sick or deviant child can represent a threat or blow to the parent's self-concept and self-confidence (Carr & Oppé 1971, Roos 1963, Schild 1979, Solnit & Stark 1961), creating parental feelings of shame, guilt, depression and uncertainty/insecurity. The emotional stress an ill child represents can have negative consequences on the parent's own life. While the child's illness can in some cases improve the marital relationship (to take care of and raise the child represents a mutual responsibility for both parents) (Drotar et al 1975, Gath 1977, Mattsson & Gross 1966), the marital relationship is often worsened, sometimes resulting in separation and divorce (Firth et al 1983, Gath 1977, Mattsson & Gross 1966, Schild 1979). Illness in a child can influence the parents' desire to have more children, often because of guilt feelings about having created a sick child; this is especially likely if the illness is genetically determined (Schild 1979).

A psychic trauma disturbs the individual's mental balance and functioning; psychological defense mechanisms are used to avoid painful and strong feelings that threaten the individual's psychic balance. These defense mechanisms are often necessary and purposeful for the individual's psychological working-through and integration of the psychic trauma, but if the defense mechanisms are very pronounced they can hinder the individual from an appropriate adaptation to the changed conditions that the trauma has created.

Among the many different defense mechanisms available, two (denial, repression) are very often observed in this context: Manifestations of denial can often be seen in the individual's not admitting that unpleasant and threatening facts about the child's illness are of importance (Chodoff et al 1964, Drotar et al 1975, Mattsson & Agle 1972, Poznanski 1973, Schild 1979). Drake and Ober (1963) have shown that parents' rep-

ression can be extensive: In a follow-up of parents of 132 children with manifest or suspected brain damage, 66 % of the parents reported that they had never been informed of the damage, in spite of medical record information indicating that they had been so informed. Parents' receptiveness to the information and their capacity to develop a realistic conceptualization of the child's illness can be influenced by the parents' emotional reactions. Parents of an ill child often have an incorrect or incomplete conceptualization of the child's illness (Leonard et al 1972, Sibinga & Friedman 1971, Wood et al 1967), which can lead to inadequate care and child-rearing.

The results and observations presented in the above literature regarding psychological and psychosocial effects on parents and families resulting from children's illness and handicaps provided the basis for constructing hypotheses and choosing topics of relevance for the study.

METHODOLOGY

The total study's design and methodology are described in detail in Project Report Part 1 (McNeil et al 1981) and summarized in article (I). A description of the design and methodology that is directly relevant to this dissertation is presented below.

Design

The study's design can be described in terms of the following questions: 1. What took place in association with the identification and follow-up of the child's ATD? To study this question, each parent with a child with ATD was interviewed concerning the events, reactions and actions in association with the identification and follow-up of the ATD. Information from medical records was also used, where possible.

2. What are the parent's current attitudes, feelings, knowledge and emotional adjustment in relation to the ATD-matter? This question was studied through parental interviews systematically judged for a number of relevant standardized variables. The interviewer also performed clinical judgments of these characteristics.

3. What other long-term consequences has the ATD-matter had for the parents, the parent-child relationship and the child? This question was studied by comparison of the ATD-group with two different control groups concerning the hypothesized consequences of the ATD-matter, defined prior

to the study. Parental interviews that were evaluated blindly per a standardized scoring system were used to study potentially relevant phenomena that normally exist in varying degrees for parents in both ATD- and matched control groups, thereby making a comparison between these groups possible.

Two control groups were used: 1. A control group pairwise matched with the ATD-group on demographic characteristics. The matching was done so that possible demographic differences between the groups would not influence the variables being studied. 2. A partially matched control group. The demographic characteristics (marital status, reproduction, social class level) that were hypothesized to have been influenced by the ATD-matter and that represented matching criteria for the matched control cases could thus not be studied through comparison with the matched control group. Another control group (partially matched controls) was thus chosen for studying the effect on demographic characteristics.

4. How can renewed screening for ATD be improved psychologically? This question was studied by investigating the ATD-group parents' positive and negative experiences in the 1972-74 ATD-screening and their recommendations regarding renewed screening for ATD.

Samples

A. The ATD-sample. The sample was to consist of a representative selection of clinically healthy children with severe ATD (PiZ form) identified through the neonatal screening in 1972-1974.

Selection criteria. Among the 120 PiZ cases identified by the screening, the cases chosen were those a/ who lived within a contiguous geographical area south of (and including) a line from Göteborg to Stockholm, b/ who had not had neonatal liver disease of clinical relevance as a result of ATD, c/ who did not have serious malformations or chronic diseases diagnosed in the neonatal period, d/ whose family was Swedish-speaking and e/ whose family had, according to medical record information, been contacted by the medical system in association with the identification of the child's ATD.

Sixty-two cases met the above criteria, while 17 did not (14 cases had neonatal liver disease, one case had chondrodystrophic dwarfism, one case had not been contacted by the medical system, and one family was not Swedish-speaking).

Sixty-one of the 62 cases (98 %) agreed to participate in the study.

No contact was obtained with the remaining case (letters were not answered and no telephone was available). The ATD-sample consisted of 37 (61 %) boys and 24 girls. The children's age at study ranged from 4 years 6 months to 7 years 1 month. Mean age was 5 years 9 months.

The total number of parents in the ATD-group at follow-up consisted of 60 mothers and 55 fathers. In 87 % of the cases, both a mother and a father were found in the home, while in a few cases there was a mother alone (10 %), a father alone (2 %) or shared custody (2 %). The parents were currently married in 74 % of the cases. All except three of the current parents were the child's biological parents. At follow-up, 88 % of the mothers and 85 % of the fathers were between 25 and 39 years of age. The group's remaining demographic characteristics are shown in Table 1 (I).

Interview data were obtained with 59 (98 %) of the mothers and 48 (87 %) of the fathers. The only mother who was not interviewed was ill at the time of the study. Among the seven fathers who were not interviewed, two did not wish to participate in the study, and the other five were not at home at the time of the study due to their work. The loss of these seven fathers did not appear to have biassed the results (Thelin et al 1982).

B. The matched control group. The goal in choosing this control group was to obtain a group that was demographically similar to the ATD-group but that did not have ATD or any chronic somatic illness whose psychological effect on the parents might be similar to that hypothesized for ATD.

Selection criteria. For each ATD-case that participated in the study, one control case was chosen and matched with the ATD-case on the following characteristics: a/ child registered at the same well-baby clinic, b/ child free from chronic somatic disease, c/ child's family Swedish-speaking, d/ child born as close in time as possible to the ATD-child, given similarity to the ATD-case on e/ child's sex, f/ social class level, g/ number of parents in the home, h/ number of siblings and order among siblings, and i/ type of dwelling (single/multiple-family).

The selection was performed on the basis of information obtained from well-baby clinic records and population register information. Sixty-one matched controls were thus chosen. Four (7 %) of the initially chosen cases did not wish to participate in the study, and they were replaced by four new matched cases. Considerable similarity was found between the ATD-group and the matched control group on all matching criteria except

social class level, which was somewhat lower in the matched control group (I). The demographic characteristics of the ATD-group and the matched control group are shown in Table 1 (I).

Interview data were obtained with all 61 mothers and 45 (85 %) of the 53 fathers in the control group. Two of the eight fathers who were not interviewed did not wish to participate, while the remaining six were not at home at the time of the study because of their work. No differences were noted in social class or other characteristics between the nonparticipating and the remaining fathers in the control group, and the attrition was judged not to have systematically influenced the results of the study.

C. The partially matched control group. The possible effects of the ATD-matter on demographic characteristics (e.g. social class, reproduction, marital status) could not be studied through use of the demographically-similar matched control group; another control group was therefore chosen.

Selection criteria. For each ATD-case participating in the study, three control cases were chosen according to the following criteria: the children a/ of the same sex, b/ from Swedish-speaking families, c/ born nearest in time, d/ registered at the same well-baby clinic as the ATD-child and e/ free from chronic somatic illness. The partially matched control group thus consisted of 183 cases.

Methods

Methods relevant for the total study. A major part of the data used in the study were obtained through visits to the homes of the ATD- and matched control cases. Home visits were thought to provide the best opportunity for motivating the children and parents to participate in the study. The children and parents could also be observed in their own natural, everyday environment.

The home visits were conducted in a parellel manner for the ATD- and matched control cases, geographical area by area, from February 1979 to March 1980.

The methods used in the total study were the following:

1. Semistructured interview with each parent separately. Each parent in the ATD- and matched control group was interviewed about different topics concerning themselves and the child related to the hypothesized long-term effects of the ATD-matter and background factors that might influence these effects. ATD-group parents were also interviewed in detail

concerning the ATD-matter and their reactions to it. Each interview took about one hour and was tape recorded for later systematic scoring by another researcher.

2. Mother-child interaction. During a 10-minute period, the mother and child were to work together to furnish a model play house and plan a day's menu for its "residents". The interaction was observed by the interviewer and also tape recorded for later blind scoring. Interaction was studied for both ATD- and matched control groups.

3. Temperament questionnaires. Standardized questionnaires regarding the child's temperament characteristics were given to all parents in the ATD- and matched control groups at the home visits. The parents were requested to complete the questionnaires independently of one another and return them to the researchers in postage-paid envelopes.

4. Clinical assessment of the parents and child. The ATD- and matched control cases were individually evaluated by the interviewer on the basis of the total home visit and also by another clinician (E. Aspegren-Jansson) on the basis of the interview transcripts. The degree to which the case showed apparent disturbance versus adequate functioning at an individual and familial level was clinically judged, as was the impact of both the ATD-matter (on ATD-cases) and other psychologically-similar factors (on both ATD- and control cases).

5. Medical record information. All medical and well-baby clinic records for the child's first 5 years of life were systematically assessed by a pediatrician (T. Sveger) for the frequency of visits to medical and health services, the reasons for the contact and the child's physical condition at these visits. This was studied for the ATD- and matched control groups.

6. Population register information. Demographic data for the parents and families were obtained from the population registers for the ATD-group and both control groups. These data represented the basis for matching the control groups and studying the possible effects on the families' demographic characteristics.

Data from methods 1 and 4-6 were used in the articles in this dissertation, as described below.

Pretesting of methods. Valuable experience with the procedure for contacting the families, with the interview technique and content, and with the order among the different methods was obtained through pretesting the total methodology on seven families of children with ATD of the less

serious PiSZ type, also identified in the same neonatal screening.

Subject contact, home visits and data collection. About two weeks before the planned home visit, the researchers sent the family a letter (that was identical for both ATD- and matched control cases, see Appendix A), which briefly and in rather general terms informed them of the purpose of the study and that they would soon be contacted by telephone to discuss a date and time for the home visit, which usually took place in the evening. At the home visit, the parents were first informed regarding the purpose of the study and the procedures for the visit, after which each parent was interviewed separately, and the mother-child interaction study was performed. The interaction was always studied after the mother had been interviewed, partially so that the child's possible shyness would have diminished and partially so that the mother would have had a chance to develop a relationship to the interviewer, establishing the necessary conditions for a natural and representative mother-child interaction. The intended order of topics in the semi-structured interview was followed as far as possible.

Specific methods. The methods used for obtaining the results presented in this dissertation are presented below.

1. Semistructured interview. An important goal in using the semistructured interview was to obtain systematic information about the parents themselves and the children in both the ATD- and control groups. The interview topics and questions were the same for both groups, in order to provide a basis for comparing them. The parents in the ATD-group were also interviewed about topics related to the child's ATD.

The order among the interview topics was the following for both ATD- and control groups: a/ basic facts about the family (number of members, age, marital status, amount and satisfaction with work, child care facilities etc.). b/ parent's description of the child (its development and personal characteristics, their attitudes toward the child, relationship between siblings, separation from parents, problem areas in everyday life etc). c/ the parent's view of the child's health (present and previous), child-related contacts with the medical system, experience of these contacts etc. d/ the parent's experience of his/her own health (mental and physical, present and previous, etc.). e/ the parent's view of his/her own life situation (current and future, view of the child's future, plans for more children, etc.). f/ for mothers only: the pregnancy and delivery of the child in question.

Interviews with ATD-group parents also contained questions concerning different aspects of the child's ATD. This was discussed in association with the above questions about "the child's health" and concerned the following topics: a/ the conditions surrounding the first news of the child's ATD (when, who, how, what was told, the parent's reactions and thoughts, etc.). b/ the continued contacts with the medical system concerning the ATD (the parent's reactions and attitudes toward the doctor's appointments, physical examinations, blood tests, etc.). c/ the parent's current knowledge, feelings, attitudes, actions etc. regarding the ATD). d/ the parent's recommendations regarding how to conduct new screening for ATD.

The interview content that was common to the parents in both groups was termed "the regular interview", while the interview content that concerned the children's ATD (i.e. content that existed only for ATD-group parents) was termed "the ATD-interview".

Editing of the interview transcripts. Each interview was tape recorded, transcribed by a secretary and the transcript edited. The purpose of this editing was a/ to divide up the transcripts for ATD-group parents into two separate parts, i.e. the content for the "regular interview" and the content for the "ATD-interview", and b/ to give the regular interview transcripts a similar appearance in both ATD- and control groups, in order to permit blind scoring of the contents.

As the first step in this procedure, the interviewer read the transcripts for both groups and removed the names of the child, doctors, hospitals and places; this was done so that the blind scorer would not be able to identify specific cases in the independent scoring of the different interview parts (both mothers and fathers) that was to follow.

As the second step, each transcript for an ATD-group parent was divided up into the regular interview and the ATD-interview. This editing was easily done, as the information about ATD was generally concentrated in one place in the interview. (In a few cases, ATD-group parents referred to the ATD-matter while discussing topics belonging to the regular interview, and these references were temporarily removed from the case's regular interview transcript in order to maintain the scorer's blindness; these parts were scored at a later time and the data used in the study. This procedure had little effect on the primary scoring; of the approximately 14,000 scores obtained for the regular interviews, only 30 were later changed due to this temporarily removed material.)

The regular interview transcripts for the ATD-group showed visual evi-

dence of removal of interview contents (cut marks), and, as this would divulge the status of ATD-group parents, a third editing step was performed: No information was removed, but the transcripts of control group parents were "edited" (i.e. cut up and photocopied) to give them an external appearance identical to that for the transcripts of ATD-group parents; the amount of apparent disruption was done on an ATD/control pairwise basis. The regular interviews for both ATD- and control group parents could thus be blindly scored.

Scoring and inter-judge reliability. Scoring systems with satisfactory inter-scorer reliability were developed both for the regular interview and for the ATD-interview. The regular interview was scored for 136 variables of which 91 (66 %) represented 5-point rating scales (e.g. from positive to negative, or much to little) and the remaining 45 represented category scales (see examples in Appendix B). On 20 cases randomly chosen from the combined samples, two blind scorers (E.A.J. & T. McNeil) independently reached an average inter-scorer correlation (Pearson product moment correlation) of + 0.80 on the 91 rating scales. For 98 % of the category scales tested, the two scorers reached perfect agreement on 80 % or more of the 20 cases' scores.

The ATD-interview was scored for 219 variables, of which 176 were scored on category scales; of the 167 inter-scorer tested, 90 % had perfect agreement on 15 (75 %) or more of the 20 cases tested. The remaining 43 variables were scored on 5-point rating scales, with an average inter-scorer agreement of r = + 0.79 for the 20 cases.

Only some of these variables were used in the articles in this dissertation.

Scoring of the interviews was done systematically in the following order: all mothers' regular interviews first, followed by fathers' regular interviews, followed by mothers' ATD-interviews, and finally fathers' ATD-interviews. All interview transcripts were scored by one and the same researcher, and each transcript was scored independently of all other transcripts and project data for the same case.

2. Medical record information. Pediatric medical and health service records for all 61 ATD-cases were obtained and studied for the number and dates of medical check-ups.

3. Clinical assessment of families at home visit. On the basis of the interviews and observations, a number of characteristics of the parents, the parent-child relationship and the child were judged by the

interviewer, and degree of adjustment to the ATD-matter was also assessed for the ATD-group parents.

4. Population register information. Information was obtained from population registers concerning the parents' occupations, place of residence and marital status. This was scored by one and the same person.

5. Statistical methods. Nonparametric statistical methods were generally used: Wilcoxon matched-pairs signed-ranks test was used for comparison of ATD- and control groups on the 5-point rating scales. Chi-square and Fisher exact P were used for analysis of category scale data (Siegel 1956). Pearson product-moment correlation coefficient was used to express inter-scorer agreement on the rating scales.

Summary and discussion of methodology

Question 1. How can the psychological and psychosocial effects on the parents resulting from neonatal identification of ATD be studied, 5-7 years later? (I)

The study can be methodologically divided into two parts. One part represents a retrospective study of only the families with a child with ATD. These parents were interviewed about what they were told, what they had experienced, how they had reacted etc, in association with ATD during the period from the first news of the ATD until the follow-up study.

The second part of the study represents a concurrent, cross-sectional investigation of the screening's long-term psychological and psychosocial effects on the parents and family. The ATD-group parents' current attitudes, feelings, knowledge etc. regarding the child's ATD were studied and assessed. Furthermore, the screening's hypothesized long-term effects on the parents and family were studied through comparison of the ATD-group and carefully selected control groups. The collection, scoring and analysis of data were performed systematically and, when possible, blindly with respect to the subject's group status and other project data. The reliability of the scoring was satisfactory. Different methods have been used in order to study the different possible effects of the ATD-matter on the parents and children. The results obtained with the different methods can complement and to some extent cross-validate one another.

Discussion of methodology. The study represents primarily an empirical investigation of psychological and psychosocial effects on the parents and children with ATD. Literature on the effect of children's illness on the family represented a basis for constructing hypotheses about the con-

sequences of the ATD-matter. Empirical, psychodynamic and crisis theoretical studies have been taken into account. Possible psychological effects on each family member per se (and especially the parents) have primarily been studied.

The study did not attempt to (nor could it, due to its form) investigate the psychodynamic processes within the parent or family. These phenomena and aspects are naturally of interest regarding the psychological effects of disease. The current results may be interpreted within such a system of reference.

The psychodynamic and crisis theoretical aspects of the parents' reactions and adjustment to illness in a child are well documented and frequently described in the literature. In contrast, relatively little is known concerning the long-term effects of such psychic trauma, and systematic studies of representative samples are clearly needed.

The reliability of parental reports. Many of the current results are based on parents' retrospective reports of early events and reactions, and question can be raised as to whether these reports are sufficiently accurate to provide adequate answers to the questions posed in this dissertation.

Parents' retrospective reports of children's psychomotoric development and illnesses, childrearing problems etc. have in a number of studies shown a varying degree of accuracy regarding the timing and nature of events and conditions (Cunningham & Sloper 1977, Drake & Ober 1963, Pyles et al 1935, Robbins 1963, Taft & Goldfarb 1964, Wenar 1963).

Study was made of the degree of accuracy or reliability of the ATD-group parents' reports of early events and conditions surrounding the child's ATD. Three different strategies were used (Thelin et al 1982): a/ A comparison between the parents' reports and medical record information regarding the timing of the early ATD-medical contacts showed a considerable degree of agreement; on two variables, the mothers' reports agreed with medical record information in 82 % and 96 % of the cases with comparable data, while a lower degree of agreement was found between fathers' reports and record information (53 % and 72 %).

b/ A comparison between the two parents' independent reports within the family concerning 12 different facts (e.g. how the contact was made, who attended the appointments, how the parents reacted) showed agreements ranging between 76 % and 98 % across these variables. The parents were very much in agreement about each others' initial reactions to the ATD-

matter when the parent had reported having reacted with anxiety-worry and shock. For example, among the mothers who reported they had reacted with anxiety-worry-shock, 90 % of their spouses also independently reported that the mother had reacted in this manner. The degree of agreement between the two parents concerning four time-related variables was lower (50-67 %).

c/ Each parent's memory for events and facts about the child's ATD was judged by the scorer of the interview on a 5-point scale (inter-scorer agreement = + 0.69). Seventy percent of the mothers and 38 % of the fathers were judged as having a "good" or "very good" memory for the ATD-related events.

In total, the above results suggest that the apparent degree of precision and reliability in the parents' (and especially the mothers') reports of early events and conditions was sufficient to provide answers to questions about these events and reactions associated with the ATD-matter.

Sample attrition, data collection and scoring. The sample was considered to be representative for clinically healthy children with ATD and their families, as all except one of the cases that met the selection criteria took part in the study. The nonparticipation by seven ATD-group fathers and eight control fathers was judged not to have resulted in any systematic bias in the results (McNeil et al 1981, Thelin et al 1982).

With the semistructured interview, almost all subjects' interviews could be scored for all variables, the results thus being representative for the total sample. By contrast, a case-oriented, clinical interview technique might have yielded more in-depth information for the individual case, but would on the other hand have led to unsystematic information for the entire group, and provided difficulty in separating the interview contents into the regular vs. ATD-interviews, thus hindering blind scoring of the interviews.

Editing of the interview transcripts appeared to have had very little effect on the primary scoring of the regular interview contents. The generally high blind inter-scorer reliability obtained suggests that the variables represented reliable dimensions on which to score the interview contents.

Children's age at follow-up and its possible effect on results. The ATD-children's age varied from 4 years 6 months to 7 years 1 month, giving a range of 2.5 years (89 % were 5-6 years old). As ATD- and control child-

ren were matched for age, this variance in age did not affect ATD-control comparisons. However, the passage of different amounts of time since ATD-identification could potentially have influenced parental reports of their early reactions and also their adjustment to the child's ATD.

To determine whether age at study was systematically related to the results of the ATD-interview, the ATD-sample was divided into a younger group and an older group, and these groups were compared with each other on parents' reports of their initial reactions (type and strength), the ATD-matter's current emotional importance for the parent, and the parents' emotional adjustment to the ATD-matter at follow-up. No differences between these two groups were found on these variables, indicating that the child's age at follow-up was not systematically related to these important effects of the ATD-matter.

RESULTS AND DISCUSSION

Results and discussion are presented in two sections below. The results based on the ATD-interviews are presented and discussed first, followed by results and discussion of comparison of the ATD-group and the two different control groups.

Results based on ATD-group parents' reports about the ATD-matter

Question 2. How were the parents first contacted about the child's ATD and how did they react to this contact? (II) Information in the medical records and the parents' reports at the interview indicated that the majority of the 61 families had been contacted before the child was 6 months old by a physician who informed the mother about the child's ATD on the telephone. Most of the parents felt that they had received unclear and insufficient information, and a majority initially conceived of ATD as representing an immediate, serious threat to the child's health (for about one third of the mothers and 10 % of the fathers it was comparable to a deadly illness). Most of the mothers (78 %) and many fathers (58 %) reported that they had reacted emotionally negatively to the news, most often with worry-anxiety-fear. The reactions were often strong (among mothers) and long-lasting.

Question 3. How did the parents experience the subsequent contacts with the medical system concerning the child's ATD? (III) The ATD-children and their parents attended several planned check-ups with pediatricians concerning the ATD during the first 5 years of life. About one

third reported that they had felt relieved after the first ATD-doctor's appointment, which typically took place about 2 weeks after they had received the first news of the ATD. This period of waiting was reported as a painful experience by almost half of the mothers. The parents' attitudes toward the special doctor's appointments about ATD varied considerably within the sample, and these attitudes were related to the doctor's apparent competence regarding the ATD and the doctor's ability to give emotional support, especially to the mothers. Most of the parents were predominantly positive and few were predominantly negative toward the child's ATD having been identified in the neonatal period, but most parents experienced the repeated blood tests for the child's liver function as especially difficult.

Question 4. What current feelings and degree of emotional adjustment toward the child's ATD did the parents show 5-7 years after its identification? (IV) Judgments of the parents' feelings and adjustment were done by the interviewer in association with the home visit and independently by a psychologist who systematically scored the interview transcripts for standardized variables. A considerable degree of agreement existed between the independent judgments of these two researchers.

At follow-up, 58 % of the mothers and 44 % of the fathers had primarily negative feelings (especially worry, anxiety, fear, guilt) toward the child's ATD. About half of the mothers and one third of the fathers were judged to have a poor emotional adjustment to the child's ATD (based on the nature, strength and manner of expression of their feelings). A considerable degree of continuity was found for the individual mother's feelings from the time of the initial contact to the time of follow-up, but continuity was lower for the individual father's feelings.

Question 5. What recommendations did the parents have concerning a new screening for ATD? (III) Most of the parents felt that ATD should be identified as early as possible in the child's life. The parents should be informed at a specially arranged doctor's appointment, which both parents should attend. Written information about ATD should be given to the parents, and they should be offered repeated doctor's appointments, preferably with the same doctor. If screening for ATD were to be reinstated, all parents should be informed in advance about the existence and methods of the screening and ATD's nature and consequences.

Discussion of Questions 2-5. The parents' reports of events and reactions associated with the first contact by the medical system regar-

ding the child's ATD concern events that took place 5-7 years earlier. In light of this time perspective, the negative reactions reported by 78 % of the mothers and 58 % of the fathers to the first news of the child's ATD should, if anything, be viewed as minimum estimates, as some memories of negative reactions may have diminished over the years.

Many of the parents' (and especially the mothers') initial negative emotional reactions were strong and long-lasting, and one third of the mothers initially thought that the child was suffering from an imminently deadly or severely handicapping disease. These findings have several implications: a/ The pediatricians' observations of individual parents' negative emotional reactions to the ATD (which strongly contributed to the discontinuation of ATD-screening after 1974) were generally confirmed for a representative sample of cases identified in the screening.

b/ The parents' reactions to the news of the child's ATD were similar to reactions to the identification of chronic somatic illness in children, extensively described in the literature (McNeil & Thelin 1980). The results thus confirm the assumption that the literature about the psychological effects of illness in children was relevant to planning the current study and constructing hypotheses for it (McNeil et al 1981).

c/ Strong anxiety and worry in the parents (and especially the mothers) associated with the idea that the newborn has a deadly disease can result in lasting psychological and psychosocial adjustment problems for the parent. Green and Solnit (1964) have termed this phenomenon "the vulnerable child syndrome", and similar phenomena have also been observed and studied by other researchers (Rose et al 1960, Rose 1961, Sigal et al 1973). In these cases, the mothers experienced fear and worry especially about the child, who in turn was at risk for psychological damage. The doctor's reassurance that the child was healthy was observed to have little effect on the mothers' continued worry and fear (Rose et al 1960).

A healthy newborn child bears symbolic importance for the mother's self-concept and identity. During the child's first months of life, the mother is especially sensitive to any deviation in the child, especially if it represents a deadly condition.

In total, many of the mothers' reactions to the news of the child's ATD and their conceptions of ATD's consequences for the child thus appear to provide the potential for long-lasting negative psychological and psychosocial consequences for the mother and child.

d/ The mere <u>suspicion</u> of a deviation or illness in the child during

its first months of life can result in lasting negative psychological consequences for some parents. Studies of the psychological effects of falsely positive screening results in neonatal screening for hypothyroidism (Bodegård et al 1983) and diverse congenital metabolic disorders (Sorenson et al 1984) showed that many parents initially reacted strongly, and in many cases (20 %) (Bodegård et al 1983) the parents had psychological problems lasting as long as a year afterward as a result of the falsely positive screening results.

At follow-up in the current study when the children were 5-7 years old, about half of the mothers and one third of the fathers evidenced negative feelings toward the child's ATD, and the same proportion of mothers and fathers were independently judged to have a "poor" or, in a few cases (8 % mothers, 6 % fathers), a "very poor" emotional adjustment to the child's ATD, with varying effects on the parent's daily life and relationship to the child. These results represent evidence of negative, long-term psychological effects on the parent resulting from the identification of the child's ATD.

A question of special interest concerns what parental and familial conditions are associated with the parent's initial reactions and long-term adjustment to the child's ATD. Increased knowledge about these conditions could enable early identification of those parents and families that are at especially high risk for negative psychological consequences of the shocking news of the child's deviation. Such special risk families could be selectively offered therapeutic and supportive contacts. Systematic analyses for the purpose of providing answers to this question are planned (I).

The parents' per se natural negative emotional reactions to the news of the child's deviation varied considerably in strength and duration within the sample. About 40 % of the mothers and 30 % of the fathers reported that the first scheduled doctor's appointment helped them to get over their initial reaction to the news of the ATD and feel calmer and somewhat relieved. The doctor's personal characteristics at the ATD-check-ups appear to have been of great importance, especially for many mothers. Parents who had come to handle the child's ATD well emotionally tended to report that the doctor had been calm and had taken time to discuss and explain the child's ATD. Many mothers felt it had been important that the doctor had given them emotional support, while the fathers tended more to stress the importance of the doctor's knowledge and competence regarding

ATD. On the other hand, many other parents reported that they had received poor information about the ATD, and in a number of cases the doctor had been hurried, uninterested and seemingly uninformed about ATD.

These results might suggest that improved routines associated with neonatal ATD-screening could diminish the adaptational difficulties associated with the identification of ATD; this improvement is of greatest importance for those families at special risk for poor adaptation. Information about the existence and purpose of screening could be given verbally or in written form to all parents before the screening is performed. For cases with ATD, as little time as possible should elapse between giving the parent the first news of the ATD and the special doctor's appointment arranged for informing about the ATD. The doctor should be especially interested in and knowledgeable about ATD and its consequences, parents' reactions to such news and how to deal with these reactions. These recommendations about screening and follow-up of identified cases are in accordance with and supplement those recommendations given by the parents. The above recommendations are also congruent with those principles that have been compiled on the basis of literature on pediatric and genetic counseling (McNeil & Thelin 1980).

The mothers' emotional reactions and adjustment to the child's ATD were consistently different from those of the fathers. A greater proportion of the mothers than the fathers reacted negatively to the news of the ATD, their reactions were in general stronger and longer-lasting, and they more often initially conceived of the ATD as representing an imminent, serious disease. At follow-up, the mothers more often had negative feelings and poor emotional adjustment to the child's ATD than did the fathers.

This greater effect on the mothers may be due to several factors: The mothers generally had had primary responsibility for the child and its health, including the ATD, during the child's first 5-7 years of life. (The mother was generally the parent who received the first news of the child's ATD and who had attended the doctor's appointments with the child (II, III).) The first months postpartum can have represented a more emotionally vulnerable period for mothers than for fathers. Differences in (expression of) emotionality between the sexes can also have contributed.

Results based on comparison of ATD- and control groups

Question 6. Had the ATD-matter influenced the parents' demographic

characteristics (reproduction, marital status and social class level)? (V)

Information obtained from the population registers and well-baby clinic records was used to study whether the ATD-matter had led to reduced reproduction, a decline in social class and an increase in the number of separations/divorces in the ATD-group families during this period. Such effects were not found in comparison of the ATD-group with a control group of 183 families living in the same area and with children of the same sex and age. An unexpected finding was that the ATD-families (fathers) had significantly higher social class level, raising the question of whether ATD is associated with a gene advantage.

Question 7. Had the ATD-matter negatively influenced the parents' attitudes a/ toward themselves as parents, b/ toward having more children (VI), and c/ toward their current life situation; and had it influenced their experienced mental and physical health? (VII) The 61 ATD-children and their families were compared with a matched control group on several blindly scored variables. No differences existed between the groups concerning the mothers' attitudes toward themselves as parents, while the fathers of the ATD-children showed a tendency toward having a more positive attitude toward themselves as parents. No significant differences were found between the two groups concerning the parents' attitudes toward having more children, the parents' experienced current life situation or the fathers' experienced health. ATD-group mothers (as compared with controls) reported significantly poorer experienced physical and mental health during the past year. This poorer maternal health was interpreted as a result of the mental stress due to the identification of the children's ATD.

Discussion of Questions 6-7. Some studies have found that mental stress on the parents resulting from having a sick or malformed child often results in marital disharmony. An increased frequency of divorce and separation has been found in some (Leiderman 1974, Tew et al 1974) but not all studies (Begleiter et al 1976, Cath 1978, Lansky et al 1978). Some authors (Tips & Lynch 1971) but not others (Fraser & Latour 1968) have observed reduced reproduction rates among the parents of malformed children, but families with known genetic risk for disease (Huntington's chorea) have not been found to have a lower rate of reproduction than that in the population (Erlenmeyer-Kimling 1977, Mattsson 1974).

In the current study, a majority of ATD-group parents had reacted strongly to the identification of the child's ATD, but no increased frequency of divorce or separation was observed in these families during the 5-7 years following the identification. No reduction in reproduction was noted for them either, even though a majority of the parents knew that ATD is inherited. These results do not speak against reinstatement of the ATD-screening.

The ATD-group families' social class level had not been influenced by the identification of the ATD, but, unexpectedly, their level was significantly higher than that of controls at follow-up (25 % of the ATD-group belonged to the upper class, as compared with 10 % in the control group, $P < 0.02$) (V). The family's social class was determined on the basis of the highest among the parents' occupations. Analyses within the family showed that the fathers alone accounted for the difference in social class between ATD- and control groups, and that the difference tended to have existed at the time of the child's birth. The fathers' higher social class level thus appeared not to have resulted from the identification of the child's ATD. As the fathers were not aware of their gene-carrier status before the child's ATD was identified, their higher social class level can not have resulted from psychological compensation, due to paternal feelings of shame and guilt about being carriers of a predisposition for illness (Schild 1979).

Question can be raised as to whether the fathers' higher social class level is genetically related to ATD. From a genetic perspective, one can speculate about whether the ATD-heterozygote has a selective advantage over that of the normal homozygote. This advantage for the heterozygote phenotype can possibly be manifested in a higher social class level, which in a total population would compensate for the reduced viability that the ATD-genotype represents in its homozygous form. The heterozygote's characteristics associated with the ATD-gene could promote its survival in the population (balanced polymorphism, Cavalli-Sforza & Bodmer 1971). Such phenomena have been observed in which heterozygote carriers of inherited diseases have an advantage (compared with that for normal homozygotes) in relation to the environment. In sickle-cell anemia, the heterozygote has increased resistance toward malaria compared with that of the normal homozygote (Cavalli-Sforza & Bodmer 1971); further, the healthy siblings of homozygotes with PKU have a significantly higher IQ than that of the normal population (Fuller & Shuman 1974, May 1977, Munro 1947).

The relationship between carrier status for ATD and high social class level, observed in the current study, should be investigated further in a study especially designed for the purpose of re-testing this unexpected finding and also for determining which personality characteristics might predispose ATD-gene carriers for high social class level.

Several investigations have studied the effects on the parents' health of living with a sick or handicapped child. Long-lasting symptoms of both a mental nature (e.g. anxiety, depression, grief and guilt feelings) and a physical nature have been reported in the literature (Gath 1977, Jackson 1977, Mattsson & Agle 1972, Mattsson & Gross 1966, Michaels & Schucman 1962, Schonell & Watts 1957). In the current study, ATD-group mothers reported experiencing worse mental and physical health than did the controls. This difference between maternal groups was found across the entire scale, from good to poor health. The most frequently reported mental symptoms were nervousness, restlessness, depression and sleep problems, while the most frequently described physical symptoms were chronic/repeated headache and back, stomach and joint problems. These physical symptoms were judged primarily to be of a psychosomatic nature.

In total, 36 % of ATD-group mothers and 20 % of the controls reported poor mental and/or physical health (VII). Nothing suggested that the mothers' ATD-carrier status had negatively influenced their health. The symptoms reported by the ATD-mothers were not typical of symptoms known to be caused by ATD, and ATD-group fathers (who are also ATD-gene carriers) did not report poorer health than controls.

The most likely explanation for the ATD-mothers' experience of poorer health was that it resulted from the mental stress that the child's ATD had represented. The mothers reacted more strongly than did the fathers, and an evaluation of the parents' retrospective reports showed that the ATD-identification had led to a psychic crisis reaction (defined by Moos & Tsu, 1977, p. 7) in 36 % of the mothers, as compared with 9 % of the fathers (Thelin et al 1982). The mothers' experienced poorer health at follow-up 5-7 years after the identification of the ATD was interpreted as a negative consequence of the ATD-screening, and this effect is relevant for the question of whether general screening for ATD should be reinstated.

Question may be raised as to the most appropriate time (child's age) for a general ATD-screening. That the mothers both showed considerably stronger reactions to the identification of the child's ATD than did the fathers (II, III) and evidenced lasting negative consequences (for their

health) 5-7 years later (IV, VII), suggests that the mothers are more psychically vulnerable than the fathers to news that something is wrong with the child. The postpartum period is an especially sensitive time for mothers, and they may be particularly sensitive to news of deviation in the child during its first months of life.

About a fourth (27 %) of the mothers reported experiencing that the news of the child's ATD came during an especially stressful period in their lives (Thelin et al 1982), and with few exceptions, these mothers spontaneously reported that the postpartum period itself was an especially sensitive and difficult time for them.

Screening for ATD and informing parents about the existence of ATD in their children may potentially be less psychologically damaging if done when the children are older. However, whether a different period than the neonatal period is in reality more psychologically appropriate cannot be empirically determined through the current study, and would require comparison of the results of the current study with those obtained in follow-up of screening/identification performed at another age.

The absence of statistically significant differences between the ATD- and control groups on the parents' attitudes toward themselves as parents (VI), toward their current life situation (VII) and toward having more children (VI) are also relevant to the question of renewed screening for ATD. The parents' attitudes toward having more children were not influenced by the identification of the child's ATD, in spite of their generally having known about the increased risk for ATD in subsequent children. These results concerning parental attitudes are in agreement with the project's findings that ATD-group parents had a reproduction rate similar to that of the larger partially-matched control group during the 5-7 years since the birth of the index child (V). The absence of long-term effects in these areas does not speak against renewed screening for ATD.

The ATD-group fathers' tendencies to have more positive attitudes toward themselves as fathers (as compared with those for controls) (VI), to experience greater satisfaction with their work (VII) and to work more (unpublished data) can be noted. Question can be raised as to whether the fathers' attitudes and behavior may represent a/ compensating psychological defenses against ATD's threat to their self-concept, b/ the result of a positive development of the parental role described for some other chronic conditions in children (Gath 1977, Mattsson & Gross 1966), or c/ personality characteristics associated with the ATD-gene. Question may also

be raised as to why the ATD-mothers showed none of these characteristics. These speculations concerning the above trends for ATD-fathers cannot be answered within the current study.

As judged in terms of parental smoking, the neonatal ATD-screening appears to have had little preventive efficacy thus far. A questionnaire study of all ATD-cases (PiZ) identified in the 1972-74 screening showed that 44 % of the mothers and 33 % of the fathers smoked at the time the children were 4 years of age (Sveger & Thelin 1981). The current study at 5-7 years showed similarly high rates of smoking in the mothers (30 %) and fathers (47 %) at that time, and these rates were just as high as the rates in the matched control group (unpublished data).

SUMMARY AND CONCLUSIONS

Identification of ATD during the child's first months of life caused strong worry and fear in most of the parents, and this was especially true for the mothers, many of whose reactions were long-lasting. A third of the mothers reported that they had initially conceived of ATD as representing an imminent, deadly threat to the child.

The parents' reactions to the identification of the ATD were similar to reactions of parents with chronically ill or handicapped children, as described in the literature.

Many parents' (and especially mothers') worry and fear resulting from the identification of the ATD also existed at follow-up 5-7 years later, and at that time about half of the mothers and a third of the fathers were judged to have emotional problems of varying degrees resulting from the identification of the child's ATD. During the child's first months of life, the mother may be especially psychically vulnerable, having an increased risk for lasting psychic problems resulting from information that the child is deviant. However, mothers may well also react negatively to such news received after the neonatal period.

No evidence was found of the hypothesized long-term effects of the ATD-screening on reproduction rates, social class level or marital status within the ATD-group families during the 5-7 years since the child's birth. Further, no negative effects were found on ATD-group parents' attitudes toward themselves as parents, toward having more children, toward their current life situation or - for fathers - their current experienced health.

Information about the child's ATD was experienced by the parents as varying in quality and amount, as was the manner in which they had been approached and treated by the medical system concerning the child's ATD. Increased knowledge about the factors that influence parents' reactions and adjustment to the child's ATD is important for improving the conduct of future ATD-screening from a psychological viewpoint and for identifying those ATD-families who are at especially high risk for negative psychological consequences. Therapy and support could be selectively offered to these cases. Within-ATD-group analyses for studying the above questions are planned.

A cost-benefit analysis of the psychological and psychosocial consequences resulting from the 1972-74 neonatal ATD-screening shows a predominance of "costs". The strong and often long-lasting emotional reactions (especially among mothers) resulting from the identification of the ATD and the mothers' poorer experienced health and poor emotional adjustment to the child's ATD represent the greatest "costs". That most parents were positive toward the child's ATD having been identified might be considered as a "benefit", while the absence of long-term effects on the families' demographic characteristics and the parents' attitudes toward themselves as parents, toward their current life situation and toward having more children might be considered to represent an "absence of costs", also of relevance.

In the total cost-benefit analysis of the neonatal ATD-screening, the project results obtained thus far concerning psychological consequences of screening must be relegated to the "cost" side. These results need to be weighed against the "real" somatic "benefits" of this screening and identification. That equally many parents still smoked regularly in the ATD-group as in the control group at follow-up suggests little real benefits thus far. Only follow-up in the future can indicate whether the identification of ATD has proven to be effective in improving the long-term health of these children with ATD.

The results presented in this dissertation concerning ATD-screening's psychological and psychosocial consequences speak against renewing screening for ATD in the same manner as that performed in 1972-1974. If somatic considerations indicate identification of ATD during childhood, and if there are not important somatic reasons for identifying ATD as early as the neonatal period, then renewed screening for ATD should - for psychological reasons - be conducted at a later time in children's lives.

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APPENDIX A. Introduction letter to the parents

To (the child's) parents:

In cooperation with the University of Lund and the Department of Pediatrics, Malmö General Hospital, we have begun a study among children and their families in southern and middle Sweden. The study is supported by the Medical Research Council and the Bank of Sweden Tercentenary Foundation.

(the child's name) has, like other children, attended check-ups through the medical system since the newborn period, in order to follow his development, physical health, etc. Many children have also been in contact with the medical system for other reasons.

The purpose of our study is to obtain increased knowledge about what the contact with the medical system has meant to the children and parents and how they have experienced the contact. Such knowledge is important for optimal planning of future contacts between the medical system and families.

You can be of great help by giving us your impressions and experiences - these are valuable to us. Participation in the study is voluntary.

Doctor Thomas Thelin would like to visit you in your home to discuss a number of questions.

Thomas Thelin will call you (during the following week)

Many thanks for your cooperation!

Yours truly

Thomas Thelin
M.D.

Thomas McNeil
docent

APPENDIX B. Examples of scoring system

Examples of 5-point rating scales are the following:

Item 4.a. "How does it go to leave the child with someone?

1 = very easy, never a problem, wonderful
2 = easy for the most part, seldom objects, "good"
3 = sometimes easy - sometimes hard, varies, depends on who it is, not so used to it, don't know
4 = quite difficult, is often unhappy/distressed, doesn't want to, has to be prepared carefully for it, only works with those she is acquainted with
5 = doesn't work at all, hopeless, never leave her, accepts only close friends/relatives"

Item 5.d. "Parent's experience of the child's negativism:

1 = does not matter, it's the way it should be, normal
2 = mostly goes well
3 = varying, sometimes you cope/sometimes not, irritating when you are tired
4 = don't know what to do about it, hard, difficult, tiresome
5 = hopeless, feels helpless, cannot cope with it"

An example of a category scale is the following:

Item 22.d. "Desires regarding the child's sex:

1 = wanted same sex as got
2 = made no difference
3 = wanted opposite sex as got"

ARTICLE I

PSYCHOLOGICAL FACTORS IN COST-BENEFIT ANALYSIS OF SOMATIC PREVENTION: A STUDY OF THE PSYCHOLOGICAL EFFECTS OF NEONATAL SCREENING FOR α_1-ANTITRYPSIN DEFICIENCY

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ABSTRACT. Nation-wide neonatal screening for α_1-antitrypsin deficiency (ATD) in Sweden was discontinued due to observations that identification of ATD in newborns seemed in some cases to have negative psychological effects on the parents and the parent-child relationship. A multifaceted study was developed to investigate systematically the psychological and psychosocial consequences of the identification of ATD in the neonatal period, as studied five to seven years after it took place. The study's basic goals, hypotheses, design, samples and methods are described.

One in every 1500 individuals in Sweden has a high risk for chronic obstructive lung disease in adulthood and somewhat increased risk for neonatal liver disease as a result of an inherited deficiency of α_1-antitrypsin (ATD), a blood-plasma protein (1). While ATD cannot at present be treated or cured, a considerable prophylactic effect postponing the onset of chronic lung disease is obtained by protecting the seriously antitrypsin-deficient individual from concentrated air pollutants, especially tobacco smoke (2). From economic and somatic viewpoints, general screening for ATD is

optimally conducted shortly after birth, using the same blood sample as that used for screening for PKU (3).

Nation-wide screening for ATD among all newborns (200,000) was conducted in Sweden from November, 1972 to September, 1974. During this time, 171 newborns with ATD were identified, and the genetic type and level of ATD were confirmed at an age of about 3 months (1). For cases with a type of ATD with potential clinical relevance (i.e. the homozygous PiZZ form with about 20 % of the normal level and the heterozygous PiSZ form with about 40 %), the parents were informed of the child's ATD by a physician at the local medical facility, and the children were to attend additional physical check-ups at 6 months and 1, 2 and 4 years of age.

During the course of informing the parents and conducting the follow-up of the cases, a number of pediatricians reported clinical observations that the identification of the child's ATD appeared to have considerably negative psychological consequences for the parents and parent-child relationship. The neonatal screening for ATD was discontinued, largely due to these observations. The strong need for a systematic study of the psychological and psychosocial consequences of ATD-identification at this age level was recognized. The results of such a study would be important for subsequent decisions regarding whether or not to resume general screening for ATD, and what the most appropriate forms for such screening and contact with the cases would be. (With the quickly expanding possibilities for early identification of health-risk conditions (4, 5), these topics have recently become of considerably increased general interest (6-11).)

The current project (12) was thus developed to investigate whether the "ATD-matter", i.e. a diversity of possible ATD-related influences inc-

luding the screening and identification of ATD and subsequent related events (e.g. informing the parents, continued blood tests and somatic examinations, etc.), had had negative effects on the parents and families, as studied 5-7 years after the ATD was identified. The project was also to provide information regarding the nature of the events and parental reactions associated with the identification and follow-up of the ATD, as well as the parents' current feelings, attitudes and knowledge regarding the ATD-matter. The project is thus a) a retrospective investigation of early events and reactions, and b) a concurrent investigation of long-term effects on the parent, child and family.

Professional literature available at the time was of little direct help in determining the likely psychological effects of screening for ATD or other risk conditions. (Recent publications (13, 14) have concerned psychological effects on the family of false-positive neonatal screening.) While the psychological effect on parents of physical illness in children had been extensively studied (6), relatively little systematic study had been done of the psychological consequences of general screening for manifest disease in children, and even less was known about the consequences of screening children for risk for future illness.

At the time of developing the project, the parents were thought possibly to have reacted to ATD as if it were a disease, as ATD represents a somatic deviation which was identified by doctors, which has relevance for serious illness in the future, and which led to immediately increased and repeated somatic examination of the child. However, ATD also had special characteristics with possible relevance for its psychological effect: a) Most ATD-children (and all those studied in the current sample) had not been ostensibly physically affected or sick as a result of ATD, and no

effects of clinical relevance were awaited until adulthood; b) even if preventive action could be aimed at the development of disease, no known practicable treatment existed for ATD itself; and c) for most parents ATD was a new, previously unheard of "disorder" which might be difficult to understand.

The manner in which a majority of parents would have reacted to, understood and adapted to the presence in their newborn infant of an unexpected, invisible, unknown, abstruse deficiency which poses little threat to the child's current health but implies the serious threat of chronic and even deadly disease in adulthood was unknown.

Previous studies of the psychological effect of somatic disease in children, viewed from empirical, psychodynamic and crisis theoretical perspectives (6), thus provided the basis for constructing hypotheses for the current study. Due to uncertainty about the areas in which an effect of the ATD-matter might likely be found and the forms it might take, especially when studied five years after its identification, a broad range of possible negative effects were included for study. The hypothesized effects of the ATD-matter were to a great extent to be represented and studied at a concrete, everyday level (see Methods below).

Hypotheses. The primary hypothesis was that the ATD-matter had negatively influenced a) the parent's feelings, attitudes and views concerning the child, the parent him-/herself and the parent-child relationship, b) the parent's attitude toward the medical system, c) the family's demographic characteristics (birthrate, social class, marital status), and d) the child's personal characteristics and behavior. This primary hypothesis was operationally expressed in more than 20 specific hypotheses which were to be systematically tested with the data collected in the study.

Design. The study design may be described in terms of four basic empirical questions posed:

Question 1. What took place in association with the identification and follow-up of the ATD? Question 2. What are the parent's attitudes, feelings, knowledge, adaptation, engagement etc. in relation to the ATD-matter at follow-up 5-7 years after identification of the ATD? To study these questions, parents with a child with ATD were personally interviewed concerning the course of events, parental reactions and actions in association with the identification and follow-up of the ATD. Supplementary information from medical records was also used, where possible. Data from the interviews and medical records were scored according to a detailed, standardized system.

Question 3. What were the other long-term consequences of the ATD-matter for the parents, children and families? Both experimentally- and clinically-oriented approaches were used to answer this question. In the experimental approach, the ATD-group was compared with specially chosen controls without ATD on the hypothetical long-term consequences of the ATD-matter. These characteristics were studied in both groups in a comparable manner using several standardized data-collection methods and sources of information (interview, observation, questionnaire, medical and population register records), and the data were scored blindly with respect to the subject's ATD versus control group status and independently across methods. The operational measure of the long-term effect of the ATD-matter was whether the ATD-group, significantly more than controls, showed evidence of the hypothesized consequences of the ATD-matter; the controls thus represented a baseline against which to evaluate the frequency of the hypothesized characteristics of ATD-cases. Two different

control groups were used, as described below under Samples.

In the clinical approach, judgments concerning the long-term effects of the ATD-matter on the parent and family and the question of whether events or conditions other than the ATD-matter can have influenced the characteristics of the ATD-cases (and controls) were performed separately by two different clinicians.

Question 4. What factors influenced the individual parent's reaction and adaptation to ATD's identification and existence? To answer this question, an analysis was done within the ATD-group, relating i) the parent's initial reaction and long-term adaptation to the child's ATD to ii) selected environmental, personal and situational conditions surrounding the identification and follow-up of the child's ATD. The latter data were obtained from interview and from medical and population register records.

Samples

The ATD-sample. This sample was chosen to be a representative group of (from an ATD-perspective) clinically healthy children with the most serious, homozygous PiZZ form of ATD, living in a large but sufficiently limited geographical area to allow interview by the same physician (T.T.).

The ATD-sample consisted of 61 cases a) who at sample selection lived in a geographical area in southern Sweden south of a line from (and including) Göteborg to Stockholm, b) who had not suffered from neonatal liver disease of clinical relevance as a consequence of ATD, c) who were not suffering from a serious congenital malformation diagnosed in the neonatal period, d) whose family was Swedish-speaking, and e) whose family was (per pediatric information sources) known to have been contacted by the medical system in association with the identification of the child's ATD. The 61

cases represented all but 1 of the 62 cases fulfilling the selection criteria; the remaining case refused all contact.

The sample consisted of 37 males and 24 females whose mean age at study was 5.8 years (range 4 yr. 6 mo. - 7 yr. 1 mo.); 89 % were 5-6 years of age. The basic demographic characteristics of the ATD-sample are shown in Table 1.

Matched control sample. The goal in selection of this sample was to obtain cases that were demographically comparable to the ATD-cases but without ATD or any chronic somatic disease whose psychological effect might be similar to that hypothesized for ATD. For each ATD-case participating in the study, one matched control was individually chosen as i) the child registered at the same Well-Baby Clinic, ii) who did not have a chronic somatic disease, iii) whose family was Swedish-speaking and iv) who was born nearest in time to the ATD-case, given similarity to the ATD-case on the following combination of characteristics: v) sex, vi) family social class, vii) current number of parents in the family, viii) number of siblings in the home, ix) order among siblings (only, oldest, middle, youngest) and x) type of housing (single/multiple-family dwelling). Four of the original 61 matched controls rejected participation in the study, and these were replaced by 4 new matched controls, all of whom accepted. The matched controls had a mean age of 5.9 years at study (range 4 yr. 7 mo. to 7 yrs. 0 mo.; 90 % were 5-6 years). The ATD and matched control groups were highly comparable to one another on demographic characteristics, as shown in Table 1.

Partially matched control (PMC) sample. To investigate the possible effect of the ATD-matter on familial social class, birthrate and marital status, an additional more randomly-selected control group was chosen. For

Table 1

Demographic characteristics of ATD and matched control groups at study

Characteristic	ATD group n (%)	Matched control n (%)
Family social class		
High	12 (20)	9 (15)
Middle	38 (62)	35 (57)
Low	11 (18)	17 (28)
Number of parents in home		
Mother and father	53 (87)	53 (87)
Mother alone	6 (10)	8 (13)
Father alone	1 (2)	0
Shared custody	1 (2)	0
Number of siblings in home		
None	9 (15)	8 (13)
One	34 (56)	40 (66)
Two	16 (26)	10 (16)
Three	2 (3)	3 (5)
Child's position in sibling order		
Only child	9 (15)	8 (13)
Oldest	23 (38)	24 (39)
Middle	4 (7)	7 (12)
Youngest	25 (41)	22 (36)
Dwelling type		
Single-family	48 (79)	51 (84)
Multiple-family	13 (21)	10 (16)
Type of residential area		
Metropolitan (Stockh./Malmö/Göteborg)	12 (20)	12 (20)
Cities (population > 30,000)	10 (16)	11 (18)
Towns (population 5,001-30,000)	5 (8)	5 (8)
Towns (population 501-5,000)	20 (33)	24 (39)
Countryside/village (pop. < 501)	14 (23)	9 (15)

each ATD-case, 3 PMC cases were selected as i) the children of the same sex, ii) born nearest in time to the ATD-case, iii) registered at the same Well-Baby Clinic and iv) not suffering from a chronic somatic disease. This sample of 183 PMC cases and the ATD-sample were compared only on birthrate and changes in social class and marital status since the birth of the child.

Method for study of ATD and PMC groups' demographic characteristics

The demographic data used for comparison of the ATD and PMC groups were obtained solely from the Well-Baby Clinic and population register systems, in a comparable manner for the two groups.

Methods for study of ATD and matched control groups

The ATD and matched control cases were visited in their homes by a researcher (T.T.), who interviewed each parent separately, observed mother-child interaction in a structured task situation, and gave each parent a questionnaire to complete regarding the child's temperament. The child's medical records were also studied independently.

a) Parental interview. Each parent was interviewed regarding a large number of items related to the hypothesized long-term effects of the ATD--matter and background factors that might influence these effects. The topics included basic familial characteristics and activities, parental view of the child's personal characteristics and problems, the child's health and parental experience of contact with the medical system regarding the child, the pregnancy/delivery and early life of the child, the parent's own health and experience of his/her own life situation, and the parent's view of the future regarding the child. ATD-group parents were also interviewed in detail concerning the ATD-matter and their reactions to it.

The interviews were tape-recorded and transcribed to written form, and the transcripts edited by the interviewer for later blind scoring by another researcher (E.A.J.), an experienced child psychologist. Special editing procedures were developed to separate the transcript for ATD-related topics from the transcript for topics that were common to the ATD and control groups, and to mask the ATD vs. control status of the subjects in the latter transcripts. Detailed scoring manuals were developed for several hundred specific items scored from the interviews. Independent inter-scorer agreement was tested on a series of cases randomly chosen throughout the samples.

b) Mother-child interaction in a structured play situation. The investigation of parent-child interaction gave the opportunity to study the parent-child relationship directly and to observe the child's behavior. During a 10-minute period, the mother-child pair were to work together to furnish a model play house as they desired, and thereafter plan a day's menu for the family who lived in the house. The interaction was observed by the researcher (T.T.), who subsequently described the interaction in terms of a number of standardized variables. Tape recordings of the interactions were later independently scored for selected characteristics by another researcher (B.H.), who was blind as to both the subject's group status and all other project data for the subject. Inter-scorer agreements were tested for these blind judgments.

c) Clinical assessment of the case in total. The ATD- and control cases were individually evaluated by the interviewer (T.T.) on the basis of the total home visit and also by another clinician (E.A.J.) on the basis of the interview transcripts. The degree to which the case showed apparent disturbance versus adequate functioning at an individual and

familial level was clinically judged, as was the impact of both the ATD-matter (on ATD-cases) and other psychologically-similar factors (on both ATD and control cases).

d) Parental questionnaires regarding the child's temperament. The study of temperament was related to the hypothesis of the ATD-matter's possible effect on the child's personal characteristics. Data were collected by a standardized questionnaire which represented nine temperament variables in terms of the child's behavior in specified situations (15).

e) Study of the child's medical records. To determine the extent of and reasons for usage of medical services, all available medical and Well-Baby Clinic records for the child for the period from hospital discharge after birth through 5 years of age were collected. Each entry in these records was scored by a pediatrician (T.S.) for possible diagnosis, degree of severity of illness/complaint, and treatment given. Inter-scorer agreement between two physicians was tested on a series of cases.

In summary, the breadth of topics studied and the varied nature of the methods used were intended to cover the many possible levels and forms of effect of the ATD-matter (e.g. effects on feelings, attitudes, behavior, health, interpersonal relations, and demographic/familial characteristics). Optimally, the findings obtained across these different methods can not only complement one another but also to some extent cross-validate one another, providing both breadth and solidity in the conclusions of the study. The results obtained with these methods will be reported in a series of articles to follow.

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ARTICLE II

PSYCHOLOGICAL CONSEQUENCES OF NEONATAL SCREENING FOR α_1 ANTITRYPSIN DEFICIENCY (ATD): PARENTAL REACTIONS TO THE FIRST NEWS OF THEIR INFANTS' ATD

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ABSTRACT. Medical record information and retrospective parental reports at interview indicated that the 61 families were typically first contacted about the child's ATD during its first 6 months of life, when a physician called the mother on the telephone and told her at least something about the child's ATD. Most parents felt they had received unclear or inadequate information. A majority initially conceived of ATD as representing an imminent, serious danger to the child's health. Most of the mothers (78 %) and many of the fathers (58 %) reported having immediately had negative emotional reactions, most often worry, anxiety and fear. These reactions were often long-lasting and, in mothers, typically strong.

A mother: "I took her to the well-baby clinic for a check-up. I thought she was only going to be weighed and measured and then I received the information that she had antitrypsin deficiency - and that was shocking information, and - well, we were - then it was total chaos. We were desperate. We tried to comfort each other I guess, but at the same time it felt like we couldn't do anything."

A father: "It was pretty unpleasant in fact. I was so terribly

unprepared for it. I was at home at the time - and then the telephone rang - and the doctor told about this thing. I had never heard of it. Right then at that instant I reacted in quite a shocked manner I guess."

The possibility of negative psychological and psychosocial effects on the family of identifying children at risk for future illness has become increasingly important because of the quickly expanding possibilities for early identification of risk groups (1-3). One such high-risk group consists of individuals with $\alpha_1$antitrypsin deficiency (ATD), which gives a somewhat increased predisposition for neonatal liver disease and greatly increased risk for chronic obstructive lung disease in adulthood (4).

Nation-wide screening for ATD among all newborns (200,000) was conducted in Sweden in 1972-1974, but discontinued thereafter due to observations by a number of pediatricians that the identification of the child's ATD appeared in some cases to have considerably negative psychological consequences for the parents and the parent-child relationship. The current study (5) was conducted to investigate systematically the psychological and psychosocial consequences of this neonatal identification of ATD in a representative sample, studied 5-7 years after the screening took place.

This article presents findings concerning a) the events that took place the first time the medical system contacted the parents about the child's ATD, b) the parents' reports of their emotional reactions at this first contact, c) their attitudes toward the information they received and toward the manner in which they were contacted and informed, and d) their initial conceptualizations of ATD's implications (dangerousness) for the

child's health.

MATERIAL AND METHODS

The 62 children with ATD PiZ, identified through screening 1972-74, who lived in southern Sweden, whose families had been contacted in association with the identification of the child's ATD, and who had not had liver disease, congenital malformations or chronic somatic diseases, were selected for study. The families of 61 of these agreed to participate (5).

Data concerning the current topics were obtained through both parental interviews and medical records. Information for topics b) - d) above was obtained solely through parental interviews, and is presented separately for mothers and fathers. An attempt was made to use pediatric records as the major source of information about topic a) (i.e. who contacted whom, how, when and what the parent was told at the first ATD-contact); however, in about 80 % of the cases these medical records contained little if any relevant information about these events and conditions, other than the fact that the family had been contacted about the child's ATD and that the child had attended ATD-check-ups on given dates. Data regarding topic a) were thus compiled on a case basis, integrating information from the medical records and both parents' interviews.

The investigator (T.T.) conducted an interview in the home with each parent separately; 59 (98 %) of the 60 mothers and 48 (87 %) of the 55 fathers in these families were interviewed regarding these topics. The loss of the few nonparticipating parents does not appear to have biassed these results (6).

The interviews were tape recorded and transcribed to written form for systematic scoring by another researcher (E.A.J.), a child psychologist

with long clinical experience. The interviews were scored for a large number of items, each represented by a detailed scoring system, with established inter-scorer reliability (E.A.J. & T.M.) (6).

The interview variables used in this article (and the inter-scorer agreements) are the following: Who was contacted (100 % identical scores on category scale), By whom (100 %), How (100 %), Amount/what the parent was informed (65 %), Attitude toward manner of contact ($\underline{r}$ = + 0.95 on 5-point rating scale), Attitude toward information (+ 0.89), Parent reacted emotionally (85 %), Reacted with Worry-anxiety-fear (75 %)/Shock (65 %)/Anger (90 %)/Sadness-sorrow-depression (90 %)/Uncertainty-wondering (65 %)/Satisfaction (85 %)/Calmness-composure (not tested), Reaction began when (95 %), Reaction strength (+ 0.94), Reaction duration (80 %), and Parent's initial conceptualization of ATD's consequences for child (+ 0.91). The reported reactions were further classified as negative (i.e. worry-anxiety-fear, shock, sadness-sorrow-depression and/or anger) or non-negative only (i.e. satisfaction, calmness-composure and/or uncertainty-wondering), as viewed from the parent's experiential perspective.

Analyses of the veridicality of the parents' reports of the early events were undertaken, by comparing the reports both within the family and with existing medical record information (6). As far as could be determined, these parental reports appeared to have a considerable degree of accuracy, representing at least an approximation to the real course of events, but nevertheless as experienced and reported retrospectively from the parent's own personal perspective (6).

RESULTS

The events at the first ATD-contact. The "first ATD-contact" was defined

as the very first occasion on which the medical system was in contact with one or both of the parents about the child's ATD. In 90 % of the cases, this contact had been made by the time the child was 4 months old (5 % were contacted at 5-6 months and 5 % between 7 months and 2 years of age). This contact was typically made by telephone (62%) or letter (18 %) or at a pediatric or well-baby clinic visit (11 %). In most cases, it was a physician (84 %) (nurse 11 %, medical secretary 2 %) who contacted the mother (87 %) (3 % fathers, 5 % mother & father, 2 % other). (In 5 cases (8 %), it was impossible to determine how they were contacted and what they were told, and in 2 cases who contacted whom.)

A majority of the families (72 %) reported having received at least some information about the child's ATD at that time, e.g. "that the child had too little enzymes in its blood, and should come to a check-up", "that there was a deficiency in a substance in her blood, she shouldn't start to smoke or be in polluted air" or "that she had a deficiency that could damage her lungs and that it was inherited from the parents".

In 20 % of the cases, the parents reported not being told about the child's ATD at that time, but were just asked to bring the child to a medical check-up. An important finding was that the emotional reactions reported by this latter group of parents were highly comparable (in both type and strength) to the reactions among parents who reported having been informed that the child had ATD (6).

Parental attitudes toward the first ATD-contact (Table 1). Almost half of both mothers (46 %) and fathers (43 %) were negative and few (15 % mothers, 7 % fathers) were positive toward the manner in which the first contact was made. Even more of the parents were negative (69 % of both mothers and fathers) toward the quality/amount of information received

at the first contact.

About one-fourth of all mothers and fathers spontaneously reported that unclear or inadequate information about ATD received at that time contributed very strongly to their emotional reactions to the first contact. Further, many of the parents observed that the medical personnel themselves did not appear to understand ATD; e.g. "The doctor didn't know what it was" and "If they are going to call and tell you about it, somebody should know what it is - she could not explain it".

Parental emotional reactions at first ATD-contact

The reported reactions were classified in the following manner: 78 % of the mothers and 58 % of the fathers had a negative emotional reaction (e.g. worry-anxiety, shock, sorrow), while 14 % mothers and 29 % fathers had a non-negative reaction only (e.g. satisfaction, uncertainty-wondering). A few mothers (7 %) and fathers (8 %) specifically reported not having reacted emotionally, and it was unclear whether the remaining 2 % mothers and 4 % fathers had reacted. All reactions that took place were reported to have begun immediately.

The frequency of the specific emotional reaction types is shown in Table 1. Worry-anxiety-fear was by far the most common specific reaction type reported by both mothers and fathers, while shock was also common among mothers. About half of the mothers and fathers reported reacting with "uncertainty-wondering", and such a reaction may have cognitive as well as emotional components. Uncertainty-wondering occurred most frequently (68 % of the time) in combination with negative reactions, and to a much lesser extent as the only reported reaction or in combination with other non-negative reactions only.

Strength and duration of negative reactions. Among the mothers who reac-

Table 1

Parental attitudes and emotional reactions at first ATD-contact

Variable	Mothers n (%)	Fathers n (%)
Attitude toward manner of contact		
Very positive	1 (2)	0
Predominantly positive	7 (13)	2 (7)
Neutral, mixed	20 (38)	14 (50)
Predominantly negative	11 (21)	6 (21)
Very negative	13 (25)	6 (21)
Attitude toward information		
Very positive	0	0
Predominantly positive	5 (9)	0
Neutral, mixed	12 (22)	10 (31)
Predominantly negative	26 (47)	12 (38)
Very negative	12 (22)	10 (31)
Specific emotional reaction type		
Worry-anxiety-fear	45 (83)	25 (60)
Shock	28 (52)	4 (10)
Sadness-sorrow-depression	7 (13)	7 (17)
Anger	1 (2)	1 (2)
Uncertainty-wondering	23 (43)	21 (50)
Satisfaction	1 (2)	5 (12)
Calmness-composure	3 (6)	1 (2)
Strength of negative reaction		
Very weak	0	1 (4)
Weak	2 (5)	2 (9)
Medium	4 (9)	10 (43)
Strong	10 (23)	6 (26)
Very strong	27 (63)	4 (17)
Duration of negative reaction		
A short while	1 (3)	0
A few days	3 (8)	1 (6)
A few weeks	13 (33)	6 (38)
Up to a year	11 (28)	2 (13)
More than a year	4 (10)	5 (31)
Up to present time	8 (20)	2 (13)

ted negatively, 86 % were judged to have reacted "strongly" or "very strongly", while less than half (43 %) of the fathers who reacted negatively were judged to have had such a strong emotional reaction (Table 1).

Examples of reactions judged as strong-very strong are the following:

A mother: "The doctor called me at home and told me about it. I was completely beside myself. I got terribly upset and he said that the child wasn't like other children. That idea stayed with me at least half a year. After that I started - I thought about it a lot then but later I started to think about it less."

A mother: "The nurse called and said that the child had an appointment for a blood test. I asked her what it was all about. She didn't know what she should say. She called to the doctor, so I got to talk with him, but I was too distraught, so I didn't even hear what he said. I was a complete wreck by the time my husband got home. I thought it felt terribly hard at the time."

The duration of the negative reactions varied considerably within the sample: Very few parents with information regarding duration reported the reaction to have lasted only a few days, while 60 % of the mothers and 50 % of the fathers with data reported the reaction to have lasted from a few weeks up to 1 year. Further, a substantial number of the mothers (30 %) and fathers (44 %) who reacted negatively reported the reaction to have lasted more than a year (in 10 parents it had lasted until the time of follow-up).

Parents' initial conceptualization of ATD's implications for the child's health. The parent's reported initial conceptualization of ATD (or corresponding condition) developed in association with the first contact was scored for the degree to which ATD was seen as representing a threat to

Table 2

Parents' initial conceptualizations of ATD's implications (dangerousness) for the child's health

Category	Mothers (n = 52) n (%)	Fathers (n = 39) n (%)
1 = Benign: no consequences now or in future	5 (10)	2 (5)
2 = Rather benign: possibly some problems in adulthood	6 (12)	3 (8)
3 = Dangerous in adulthood: no immediate negative consequences but definitely some in adulthood	8 (15)	11 (28)
4 = Possibly dangerous in childhood: may lead to chronic disease immediately or in near future	15 (29)	19 (49)
5 = Very dangerous in childhood: implies imminent severe chronic or lethal disease	18 (35)	4 (10)

the child's health. Each parent's conceptualization was scored according to a 5-point scale, ranging from "1 = Benign, no consequences" to "5 = Implying imminent, severe chronic or lethal disease" (Table 2). Category 3 represents the level that most closely corresponds to ATD's true consequences regarding lung disease in adulthood; Category 4 might describe the risk associated with neonatal liver disease.

Only 15 % of the mothers and 28 % of the fathers for whom this variable could be scored conceived of ATD as first representing a definite threat in adulthood (Category 3), while a small number of mothers (21 %) and fathers (13 %) initially conceived of ATD as generally benign (Categories 1 & 2).

In contrast, two-thirds of the mothers (63 %) and fathers (59 %) with data reported having initially thought that ATD had possibly serious, immediate consequences for the child's health (Categories 4 & 5). In fact, one-third (35 %) of the mothers and 10 % of the fathers initially thought that ATD implied an imminently deadly or severely handicapping chronic disease. One-fourth of the mothers (24 %) and a few fathers (6 %) mentioned initially associating ATD with cancer, PKU, mental retardation or digestive disease (6).

DISCUSSION

As determined through retrospective parental reports and medical record information, the parents were as a rule first contacted about the child's ATD before the child was 6 months old, when a physician called the mother on the telephone and told her at least something about the ATD. Exactly what each family was informed at this first contact is impossible to determine; the medical records contain little relevant information, and

the parental reports were in some cases sketchy and unclear on this point. The unclarity may be due to simple forgetting, to mental blocking of memories of this potentially threatening information or to having received unclear or inadequate information at the time. Most parents were not satisfied with the information they received, and many questioned whether the medical personnel themselves understood ATD.

Whatever was said, inferred or not said to the parent at this first contact, a majority of both the mothers and fathers initially conceived of ATD as implying an immediate, possibly serious danger to the child's health, and one-third of the mothers thought it meant an immediately deadly or severely handicapping chronic disease. The contact led to negative emotional reactions reported by almost 80 % of the mothers and 60 % of the fathers. The negative reactions were strong for most of these mothers, and lasted for at least a year in many of the mothers and fathers reacting negatively. Given the retrospective nature of these reports, the frequency of negative reactions might well represent at least minimum estimates of the negative responses that appear to have occurred at the first contact.

The mothers typically reacted more strongly negatively than did the fathers; this may be due to most mothers' having received the contact, while only about 10 % of the fathers were known to have been directly involved. Increased maternal emotional sensitivity during the postpartum period and/or possible sex differences in emotionality and in parental roles may also have contributed to this difference in reported reactions.

The emotional reactions reported by this representative sample of ATD-group parents thus confirm the pediatricians' earlier observations of individual cases (5), that the identification of the child's ATD had considerably negative psychological effects on the parents, at least ini-

tially. Parents who reported not having been told of the child's ATD at this contact reported reacting equally negatively and strongly as those who were told. This is similar to the findings in another recent study (7): Parents whose children were called to a follow-up exam after primary screening and who reported not being informed about the child's suspected disorder (i.e. were told nothing specific, told that the new test was only routine or told the first test was unreliable or lost) were found to react emotionally as strongly negatively as those parents who were informed about the child's suspected disorder. If these parental reports are correct, then any suggestion of abnormality in the child early in life carries the potential for eliciting a considerably negative emotional response in the parent.

This potential for negative effects on the parents, at least initially, must be taken into account in evaluating and planning possible future neonatal screening for ATD and other risk-conditions. Nevertheless, the assessment of the total effect of identifying risk-conditions in early childhood requires not only study of initial effects but also systematic investigation of the long-term consequences for the parents and family, including the parents' subsequent psychological adaptation to the child's risk.

Many of the parents reacted as if ATD represented manifest or imminent disease. This validates the project's use of the "psychological response to disease" model for developing hypotheses about the consequences of identifying ATD in young children (5, 8). Also, the frequently negative nature and long duration of the parents' initial emotional reactions would appear sufficient to provide a background for other potentially negative, long-term effects on the parents themselves, the parent-child relations-

hip, the child and the family in total. Such possible negative consequences, investigated in other parts of the current project (5), will be presented in subsequent articles.

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ARTICLE III

PSYCHOLOGICAL CONSEQUENCES OF NEONATAL SCREENING FOR α_1 ANTITRYPSIN DEFICIENCY (ATD): PARENTAL ATTITUDES TOWARD "ATD-CHECK-UPS" AND PARENTAL RECOMMENDATIONS REGARDING FUTURE SCREENING

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ABSTRACT. The parents of 61 children with ATD typically attended repeated doctor's appointments concerning the child's ATD during the first years of life. Many (30-40 %) of the parents felt somewhat relieved about the ATD after the first appointment. Parental attitudes toward the appointments varied considerably within the sample, being related to the physician's reported knowledgeability-understandability regarding ATD and emotional supportiveness (toward mothers). Most parents were positive and few were negative toward the child's ATD having been identified at this age. Repeated blood tests for the child's liver function were experienced negatively by most parents. The parents' recommendations concerning screening and follow-up of ATD in children are presented.

Nation-wide Swedish neonatal screening for α_1 antitrypsin deficiency (ATD) was conducted in 1972-1974, but was discontinued thereafter due to clinical observations of negative psychological consequences for the parents and families. The present study was initiated to study systematically the psychological and psychosocial consequences in a representative

sample, studied 5-7 years after the children's ATD was identified (1). Most parents reported having reacted negatively emotionally (with worry, anxiety, fear), often strongly, upon receiving the first news of the child's ATD by telephone or letter early in the child's life (2).

This article presents results concerning the families' subsequent contacts with the medical system about the child's ATD, from the time of the first scheduled doctor's appointment regarding ATD up to the current study, 5-7 years later. Approximately five different ATD-check-ups were planned for this period (3), and the local physicians were to examine the child, obtain blood samples for assessing liver function and inform the parent(s) further about the child's ATD. In this article, special consideration is given to the parents' experience of and attitudes toward these check-ups, and the parents' recommendations for how the medical system should act with respect to the identification and follow-up of ATD in children.

MATERIAL AND METHODS

The material and methods are described in greater detail elsewhere (1, 2, 4).

The 61 children with ATD PiZ, identified through screening 1972-1974, who lived in southern Sweden, who had not had liver disease, congenital malformations or chronic somatic diseases, and whose families had both been contacted in association with the identification of the child's ATD and agreed to participate in this follow-up study, constituted the present sample.

Data on the occurrence and timing of the first ATD-related doctor's appointment were obtained from the medical records. Data for the remai-

ning variables were obtained from interviews with each parent separately (59 mothers, 48 fathers). The interview transcripts were scored in a systematic manner per variables for which inter-scorer agreement was tested. The parental reports appeared to have a considerable degree of accuracy, representing the parents' own experience and memory of the course of events (5).

Two of the category-scale variables (Who attended first appointment, Family had later contact with medical system regarding ATD) are presented on a case-basis, integrating information obtained from both parents. The remaining category-scale variables are presented for each parent separately: Took action regarding ATD between first contact and first doctor's appointment; Changes in emotions regarding ATD after first appointment; How well first appointment went; How doctor was described; Parent's motives for taking part in later contacts; When should screen/inform; How should inform; Number of appointments desired; Number of parents who should be present; and Parent's spontaneous suggestions regarding ATD. Inter-scorer agreements on the above 34 category-scale items ranged from 65 % to 100 % perfect agreement on the category scale (mean = 83 %, median = 82.5 %), with 79 % of the coefficients $\geq$ 75 %.

The following additional variables were scored on 5-point rating scales ("very positive" to "very negative") for each parent: Attitude toward first appointment (inter-scorer r = + 0.85); Attitude toward amount/quality of information received at first appointment (+ 0.86); Attitude toward subsequent ATD-contacts (+ 0.86); Attitude toward medical personnel's way of acting at subsequent contacts (+ 0.87); Attitude toward child blood tests (+ 0.75); and Attitude toward having identified child's ATD (+ 0.92).

RESULTS

A. The first ATD-doctor's appointment

The first ATD-doctor's appointment was defined as the first planned occasion on which a physician formally informed the parent(s) about ATD. Approximately 2-3 weeks had typically elapsed between the first news the parent received of the child's ATD (by telephone or letter) and the first appointment. During this time, most parents (58 % mothers, 82 % fathers) took no specific actions regarding the ATD, and those who did attempted to obtain more information about ATD, from the medical system (27 % mothers, 15 % fathers), from friends and relatives (13 % mothers, 3 % fathers) and from books (4 % mothers, 3 % fathers). This period of waiting for the appointment was mentioned as an emotionally difficult time by a number of mothers (44 %) and fathers (22 %) (5).

The medical records of 59 of the 61 cases indicated that such an ATD-doctor's appointment had occurred, as did the parents' reports in the two remaining cases. In all 61 cases, one or both of the parents personally met a physician and were informed about the ATD. In 66 % of the cases the mother alone attended with the child, in 25 % both the mother and the father attended, and in 5 % she and a friend or other relative attended. In 5 % of the cases it was impossible to determine which of the parents attended this appointment.

According to medical record information, about 95 % of the appointments occurred during the children's first 6 months of life (89 % before 5 months of age, 5 % at 5-6 months and 3 % from 7 months to 2 years of age; two cases had missing data).

The first appointment's effect on parental emotional reactions to the

ATD

As all but six cases had already had their first contact with the medical system about ATD before attending the doctor's appointment, the parents' emotional reactions associated with the appointment were studied in terms of possible changes in their initial emotional reactions. (The emotional reactions for the six cases receiving their first contact regarding ATD at this appointment are included in data reported previously (2) and not here.)

Almost one half (43 %) of all mothers and one third (30 %) of all fathers reported having felt calmer and relieved about the child's ATD as a result of the first appointment. Feelings of relief were most frequent among parents who had reacted emotionally negatively to the first news of the child's ATD, but relief was also reported by some of the parents who reported only having reacted with uncertainty and wondering upon receiving the first news of the ATD.

No change in feelings was reported by 32 % of the mothers and 16 % of the fathers, while more negative feelings (worry and anxiety, anger, or sadness and depression) were reported by a few of the mothers (13 %) and fathers (9 %). The parents' reports for the remaining mothers (11 %) and fathers (44 %) did not indicate what effect, if any, the first appointment had had on their feelings toward the ATD.

Parental attitudes toward the appointment and information received

Both the mothers and the fathers reported a broad range of attitudes (from very positive to very negative) toward the first appointment in total and toward the quality and amount of information received at it (see Table 1). Given the highly varied parental attitudes toward the doctor's appointment, these attitudes were analyzed in relation to the parent's report of

Table 1. Parents' attitudes toward ATD-check-ups and identification of ATD in children

Variable	n	Parental attitude n (%)				
		Very positive	Positive	Mixed	Negative	Very negative
Attitude toward first appointment						
Mothers	56	6 (11)	11 (20)	18 (32)	14 (25)	7 (13)
Fathers	35	5 (14)	8 (23)	16 (46)	5 (14)	1 (3)
Attitude toward amount/ quality of information at first appointment						
Mothers	56	6 (11)	6 (11)	13 (23)	20 (36)	11 (20)
Fathers	38	5 (13)	10 (26)	8 (21)	11 (29)	4 (11)
Attitude toward medical personnel at later contacts						
Mothers	48	9 (19)	13 (27)	12 (25)	12 (25)	2 (4)
Fathers	33	6 (18)	8 (24)	10 (30)	7 (21)	2 (6)
Attitude toward child blood tests						
Mothers	43	0	3 (7)	7 (16)	9 (21)	24 (56)
Fathers	23	0	2 (9)	3 (13)	13 (57)	5 (22)
Attitude toward having identified child's ATD						
Mothers	46	5 (11)	20 (43)	16 (35)	4 (9)	1 (2)
Fathers	33	7 (21)	13 (39)	8 (24)	5 (15)	0

the doctor's personal characteristics and behavior at this appointment.

Appointment went well: Only 38 % of the mothers but 63 % of the fathers with data felt that the appointment had gone well. These mothers tended to describe the doctor as having been calm, interested/helpful and, less frequently, knowledgeable. The fathers who felt the appointment had gone well tended to report the doctor as knowledgeable and calm and, less often, interested/helpful. Parents who were very positive to the appointment reported e.g. that the doctor was attentive to both the parent and child, took time to explain things to the parents, showed interest in giving a good explanation of things, and attempted to reassure and support the parents emotionally.

Appointment went poorly: The 34 % of the mothers and 10 % of the fathers who felt this most often reported that the doctor was unknowledgeable or uninformed about ATD. Some mothers also mentioned that the doctor was hard to understand. Parents with very negative attitudes toward the appointment reported e.g. that the doctor appeared reluctant to inform them about ATD, did not seem to know anything about ATD, explained ATD very poorly and/or was rude or brusque with the parent. (The remaining one fourth of the mothers and the fathers felt the appointment had gone just "so-so".)

B. Further contact with the medical system regarding the child's ATD after the first ATD-doctor's appointment

A vast majority (93 %) of the cases reported having had continued contact after the first appointment. Many parents mentioned several different motives for taking part in these later contacts. The most common motive among mothers (77 %) and fathers (66 %) was that of participating for the child's sake, i.e. to contribute to the child's present and future

health. About 40 % of the parents appeared to have participated for their own sake, e.g. so as to feel more confident and at ease about the child's ATD and learn how to deal with it. A number of parents (23 % mothers, 19 % fathers) participated out of general helpfulness, e.g. to make a contribution to research. About one third of the parents (40 % mothers, 31 % fathers) participated because they felt some coercion or demand from the medical system to do so. Most (86 %) of the mothers experiencing coercion had at least one other motive for participating (usually for the child's sake), but about half (54 %) of the fathers who reported an experienced demand had no other known motive for participating.

Parents' satisfaction with later ATD-related medical contacts

The parents' attitudes toward the contacts in total correlated very highly (r = + 0.94 for mothers, + 0.97 for fathers) with their attitudes toward the medical personnel's way of acting at these contacts, and only the latter data are presented (Table 1). The parents' attitudes toward the medical personnel (all personnel mentioned) during this period was again varied, being reported as (very) positive by almost half of the parents (46 % mothers, 42 % fathers) and as (very) negative by about one fourth (29 % mothers, 27 % fathers).

Parents with especially positive experiences of the contacts mentioned that the personnel were patient and showed no irritation with the children or parents, that they had had the same doctor at different appointments, and that the doctor was understanding and affectionate and took time to discuss things with the parents. Parents with especially negative experiences mentioned that the personnel did not notice how upset and depressed the parent was because of the child's ATD and that the doctor gave no explanation for the medical procedures but only said that

"it had to do with research".

The repeated blood tests for the child's liver function were experienced negatively by three fourths of the parents (Table 1). The blood sampling technique itself was that aspect most frequently reported as negative, but the parents also mentioned the child's behavior (e.g. acting scared, screaming), the child's young age ("he was too young to subject to such tests"), the parent's separation from the child during the test ("it's hard when you hear him crying in the other room"), the personnel's unfriendly attitude toward the parents, and their general dislike for blood tests as other negative aspects.

C. Parents' recommendations regarding how the medical system should act with respect to ATD in children

The parents were asked for their views and recommendations on how the medical system should act regarding the possible identification and follow-up of ATD in children, and the parents generally answered on the basis of their own personal experience.

How does the parent feel at present about having gotten to know about the child's ATD? Should ATD be identified at all in children?

More than half of the parents (54 % mothers, 61 % fathers) stated that they were positive or very positive toward having gotten to know about the child's ATD, while a small minority (9 % mothers, 15 % fathers) were predominantly negative toward this; only one parent (a mother, 2 %) was strongly negative and reported feeling it would have been just as well not to have been informed about the child's ATD. The remaining one third of the mothers (35 %) and one fourth of the fathers (24 %) had mixed feelings or no determinable positive or negative attitude toward this identification (Table 1).

When should ATD-screening be done and the parents informed of the results?

Two thirds of the mothers (66 %) and fathers (68 %) thought that the child's age (typically 2-4 months) at which they had received the ATD-information or even earlier is the best time to inform the parents about the child's ATD. A small minority (16 % mothers, 10 % fathers) recommended that the identification and information should be done later in life (unspecified age), while about one fifth (18 % mothers, 23 % fathers) did not feel they could answer this question.

How should the medical system inform the parents about the child's ATD?

A considerable majority of both mothers (85 %) and fathers (92 %) recommended having a doctor's appointment especially arranged for the purpose of informing about the existence and nature of the child's ATD. A number of parents who recommended having such an appointment also suggested that the parents should be contacted by telephone (21 % mothers) or letter (9 % mothers, 3 % fathers) prior to this special appointment. Very few parents recommended giving information at a routine visit to the well-baby (8 % mothers) or pediatric clinic (3 % mothers), or by telephone (5 % mothers, 6 % fathers) or letter (3 % fathers).

Further, a clear majority of both mothers (80 %) and fathers (81 %) recommended having several different appointments for informing about ATD. Only one visit was seen as sufficient by 19 % of the mothers and 17 % of the fathers, while two parents had no opinion.

About three fourths of the parents (77 % mothers, 75 % fathers) thought that both parents should be present when the formal information about the child's ATD is given. A minority felt that one parent was sufficient (11 % mothers, 18 % fathers), that it did not matter how many

parents received the information (9 % mothers, 7 % fathers) or that the parents themselves should be allowed to choose whether both would be present (4 % mothers).

Additional, spontaneous suggestions mentioned by parents were that the same physician should follow-up the child's ATD for a given case (27 % mothers, 17 % fathers) and that written information about ATD should be provided for the parents of children with ATD (20 % mothers, 17 % fathers). Some parents (27 % mothers, 8 % fathers) mentioned that they had not been aware that an analysis for ATD was to be performed on the blood sample taken for PKU-screening, and three fourths of these mothers and half of these fathers expressed a desire to have known about the ATD-screening at the time it was done.

DISCUSSION

After having received the first news of the child's ATD by telephone or letter, the parents attended a number of doctor's appointments with the child concerning the ATD. The parents varied considerably in their attitudes toward the appointments, the parents' attitudes being associated with the doctor's (reported) knowledgeability-understandability and calmness-helpfulness-interest. While both mothers and fathers tended to stress the importance of the doctor's knowledgeability and understandability, the mothers also tended to stress the importance of their emotional contact with and support from the doctor.

A majority of the parents were positive and few were negative toward the child's ATD having being identified at this age; this parental attitude toward the results of the 1972-74 ATD-screening is relevant to cost-benefit analysis of neonatal ATD-screening. In contrast, the repeated

blood tests for assessing the child's liver function were generally negatively experienced by the parents (and children), and this negative effect should be weighed against the real somatic value in obtaining this information.

The parents' recommendations regarding future ATD-screening/follow-up can be summarized as indicating that - according to parents of ATD-children - (a) ATD should be identified and the parents informed early in the child's life, at (b) a specially arranged doctor's appointment, which (c) both parents attend, with (d) written information and (e) repeated appointments - preferably with (f) the same physician - for receiving more information about ATD. These parents' recommendations are generally consonant with a number of principles previously established on the basis of professional literature about pediatric and genetic counseling (6).

Further, if neonatal ATD-screening were resumed, all parents should be (g) informed in advance that analysis for ATD will also be performed on the blood sample taken for PKU. This awareness could give the parents the opportunity to prepare emotionally (i.e. emotional inoculation, 7) for the eventuality that an abnormality of that type could be identified in children at that age. Such information could be given in written form to all mothers at the prenatal clinics or at the maternity ward after delivery.

If the total cost-benefit analysis to come suggests renewed general ATD-screening in early life, then the parents need to be contacted initially to motivate them to come to the recommended special appointment. This difficult but necessary task could be done e.g. by telephone or personal contact at a routine pediatric check-up, and must be done by a physician who is well-informed about ATD and who has sufficient time and

interest to inform the parent(s) well, who can show acceptance toward and understanding of the parents' often negative initial reaction to the news, who can judge their need for additional emotional support, and who is prepared to see the entire family again soon to discuss the ATD further.

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ARTICLE IV

IDENTIFYING CHILDREN AT HIGH SOMATIC RISK: PARENTS' LONG-TERM EMOTIONAL ADJUSTMENT TO THEIR CHILDREN'S ALPHA$_1$ ANTITRYPSIN DEFICIENCY (ATD)

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ABSTRACT - The parents of 61 children at high somatic risk due to α_1 antitrypsin deficiency (ATD) were followed-up 5-7 years after the identification of the ATD and studied regarding their long-term emotional adjustment to the child's ATD. This was assessed both by a physician who interviewed the parents in their home and independently by a psychologist who systematically scored the interview transcripts for selected variables. Notable agreement was found in the independent assessments performed by these two researchers. At follow-up, 58 % of the mothers and 44 % of the fathers had predominantly negative feelings (worry, guilt) about the child's ATD. About half of the mothers and a third of the fathers were judged to have poor long-term emotional adjustment. Considerable continuity was found in mothers' feelings across the 5-7 years since identification of the ATD.

Evaluation of the psychological and psychosocial effects on the family resulting from identifying children at risk for future illness has become

increasingly important because of the quickly expanding possibilities for early identification of risk groups (1-3). One such somatic high-risk group consists of individuals with α_1 antitrypsin deficiency (ATD), which gives a somewhat increased predisposition for neonatal liver disease and greatly increased risk for chronic obstructive lung disease in adulthood (4). Development of the lung disease can be postponed considerably by protecting individuals with ATD from tobacco smoke (5).

Nation-wide screening for ATD among all newborns was conducted in Sweden in 1972-1974. The parents were typically informed of the child's ATD by a local pediatrician during the child's first months of life, and thereafter attended a number of doctor's appointments, the purpose of which was to examine the child, obtain blood samples for liver function and inform the parents further regarding the ATD (6, 7). The children with ATD (excluding those with neonatal liver disease) were not sicker than control children without ATD during the first 5 years of life (8).

Neonatal ATD-screening was not continued after 1974 due to observations by a number of pediatricians that the identification of the child's ATD appeared in some cases to have considerably negative psychological consequences for the parents and the parent-child relationship. The current study (9, 10) was conducted to investigate systematically the psychological and psychosocial consequences of this neonatal identification of ATD in a representative sample of 61 cases, studied 5-7 years after the screening took place.

At follow-up, a majority of the parents reported having initially conceived of ATD as representing an imminent, serious danger to the child's health, and a majority (78 % mothers, 58 % fathers) reported having reacted negatively emotionally (most often with worry-anxiety-fear) upon first

learning of the child's ATD (11).

The parents' initial reactions to the child's ATD were thus generally comparable to reactions previously observed among parents of children with serious illness or congenital malformations (11). Such feelings, especially if strong, can be viewed as indicating a traumatic psychic crisis for the parent (12-15), and indeed crisis reactions could be identified in the retrospective reports of a number of mothers (36 %) and fathers (9 %) (16).

In a psychic crisis, the individual's adaptive psychological mechanisms attempt to maintain a psychic balance while a working-through and integration of the trauma takes place within the individual (13, 17). Satisfactory resolution of the crisis is characterized both by the absence of mental symptoms or strongly painful feelings associated with the trauma and by the absence of negative effects of it on the individual's self-concept, family life and social functioning (13, 17, 18).

An inability, in the long run, to handle and integrate the feelings aroused by the child's illness/deviation has been viewed by many authors (14, 19-22) as the predominant factor in parents' deficient adaptation to the illness. Unresolved feelings such as worry, guilt or anxiety associated with the child's illness can not only have other negative effects on the parent's own life but also hinder the parent from developing a realistic conceptualization of the child's illness and its consequences, thereby preventing the parent from acting appropriately toward the illness and the child's special needs (20, 23-25).

Investigation of the parent's long-term emotional adjustment to the child's ATD is thus an important part of the current project's evaluation of the psychological effects of neonatal identification of ATD.

This article presents a systematic investigation of the parents' feelings and emotional adjustment in relation to the child's ATD, at the time of follow-up 5-7 years after its identification. The current feelings were also studied in relation to the parent's reported emotional reactions upon receiving the first news of the child's ATD, in order to obtain a long-term perspective for these feelings.

Different aspects of the parent's feelings and emotional adjustment to the child's ATD at follow-up were evaluated both by a researcher who conducted interviews in the home with both parents and, independently, by another researcher who systematically scored the interview transcripts for relevant variables. These two evaluations complement one another, one taking advantage of the interviewer's total clinical impressions from the home visit (including observation of mother-child interaction, 10), and the other employing greater scientific stringency (being based on detailed and independent scoring of each parent's transcript for variables with established inter-scorer agreement).

SAMPLE AND METHODS

The sample and methods are described in greater detail elsewhere (9, 10).

The ATD-sample consisted of families of 61 children with the homozygous PiZZ form of ATD, identified through screening in 1972-74, living in southern Sweden, and free from liver disease, congenital malformations and chronic somatic diseases. The children's mean age at study was 5.8 years. Interviews for the current topics were obtained with 59 (98 %) mothers and 48 (87 %) fathers; loss of the few nonparticipating fathers did not appear to have influenced the results (16).

Each parent was interviewed separately in the home by a physician

(T.T.) concerning i.a. the course of events and parental reactions associated with the identification and follow-up of the child's ATD, the parent's views and feelings regarding ATD at present, their actions regarding the ATD during recent years, etc.

Evaluation of interview transcripts. The interviews were tape recorded, transcribed and edited for systematic scoring by another researcher (E.A.J.), a child psychologist with long clinical experience. The interview transcripts used for scoring the present variables contained information directly related to the identification and follow-up of the ATD. The variables scored from the transcripts were each represented by a detailed scoring system, with established inter-scorer reliability (E.A.J. & T.M.) (9). Interviews for all mothers were scored first, followed by interviews for all fathers; both groups were scored in random order and independently within each family.

The variables scored from the transcripts and used in this report are the following (their interscorer agreements were expressed in r for the 5-point rating scales and in % perfect agreement for the 4-point category scales): Parent's current positive/negative feelings regarding the child's ATD and its consequences (r = + 0.88), The ATD's degree of emotional importance for the parent (+ 0.80), Parent's experience of personal competence regarding the ATD (+ 0.90), Interest in and need for discussing the ATD with interviewer (+ 0.79) and Specific current feelings regarding the ATD: Worry-anxiety-fear (75 %), Guilt (80 %), Sorrow-sadness-depression (90 %), Anger (90 %), Satisfaction (90 %), and Uncertainty-wondering (75 %).

Evaluation by interviewer. Immediately after each home visit, the interviewer rated each parent's Degree of emotional adjustment to the

child's ATD, weighing together impressions of the type, strength and manner of expression of the parent's current feelings and their possible effect on the parent's relationship to the child and family; emotional adjustment was scored on a 3-point scale (adequate, somewhat poor and very poor).

Statistical methods. The degree of association between the two researchers' assessments and the degree of stability of parental feelings over time were analyzed by Chi-square and Fisher exact probability (one-tailed).

RESULTS

Evaluation by scorer of interview transcripts

Parent's current feelings toward the child's ATD. At the time of the interview 5-7 years after the identification of the child's ATD, 58 % of the mothers and 44 % of the fathers were judged as having in total predominantly negative or strongly negative feelings toward the ATD and its consequences (Table 1), and most of the remaining parents had an admixture of negative and positive feelings.

With respect to the specific feelings and emotions (Table 1), a majority of the mothers and many of the fathers had feelings of worry-anxiety-fear; feelings of guilt, anger and sadness-sorrow-depression were less common. About half of the mothers and many fathers experienced uncertainty-wondering about ATD, while approximately 30 % of the parents expressed satisfaction.

The ATD's current emotional importance for the parent. The scorer judged this on the basis of the parent's manner of discussing the ATD, weighing together such components as the parent's expressed interest in

ATD, engagement with it, expressions of emotions, attitudes and possible actions with respect to the ATD. The child's ATD was judged to have "(very) strong" emotional importance for about a fifth of the mothers and fathers at present, i.e. it had taken an important place in the parent's life and view of the child (Table 1). Further, for about half of the parents, the ATD was of "some" (i.e. a moderate degree of) importance, e.g. the parent felt that the ATD was possibly important in the future but took a wait-and-see attitude at present, thought about it sometimes, etc.

Parent's experienced degree of competence regarding the child's ATD. This was judged on the basis of the parent's experienced need for more information about ATD, feelings of mastery versus uncertainty and anxiety in relation to the ATD, etc. A considerable range of scores was found for both mothers and fathers (Table 1).

More than a third of both mothers (37 %) and fathers (40 %) experienced (very) little competence regarding the ATD (e.g. they expressed uncertainty, did not know what to do at a practical level, experienced a great lack of knowledge, felt totally helpless regarding the ATD, etc.). In contrast, "very great" or "some positive degree" of experienced competence characterized about a fourth of the parents, while the remaining third of both mothers and fathers showed an uneven pattern, being confident about the ATD in some ways and insecure in others.

Parent's interest in discussing the child's ATD with the interviewer. This variable was thought to constitute one possible index of the parent's personal relationship and attitude toward the child's ATD. Approximately 40 % of the parents were judged as showing "a (great) need" to discuss the ATD, i.e. the parent expanded on the topic, took up additional aspects spontaneously, talked in detail about her/his feelings and

Table 1

Parent's feelings about the ATD at follow-up

Variable	n	Category n (%)				
Current feelings in total toward the ATD		Very positive	Positive	Mixed	Negative	Very negative
Mothers	59	0	8 (14)	17 (29)	28 (47)	6 (10)
Fathers	48	0	8 (17)	19 (40)	20 (42)	1 (2)
Emotional importance of the ATD		None	Little	Some	Strong	Very strong
Mothers	59	1 (2)	10 (17)	34 (58)	12 (20)	2 (3)
Fathers	48	4 (8)	15 (31)	21 (44)	7 (15)	1 (2)
Own experienced competence regarding the ATD		Very much	Some	Uneven	Little	Very little
Mothers	59	5 (8)	11 (19)	21 (36)	14 (24)	8 (14)
Fathers	48	2 (4)	11 (23)	16 (33)	12 (25)	7 (15)
Interest in discussing the ATD with interviewer		Active avoidance	Some avoidance	Some interest	A need to discuss	Great need
Mothers	59	0	5 (8)	31 (53)	16 (27)	7 (12)
Fathers	48	0	7 (15)	20 (42)	11 (23)	10 (21)

Specific feelings toward the ATD	Mothers (n = 59)	Fathers (n = 48)
Worry-anxiety-fear	37 (63)	18 (38)
Guilt	11 (19)	7 (15)
Anger	10 (17)	2 (4)
Sadness-sorrow-depression	5 (8)	4 (8)
Uncertainty-wondering	31 (53)	19 (40)
Satisfaction	18 (31)	13 (27)

thoughts, sought additional contact with the interviewer about ATD, etc. (Table 1). Most of the remaining parents showed moderate interest in discussing the ATD with the interviewer, and only a few were resistent toward discussing it.

To summarize across these variables, predominantly negative feelings toward the ATD were found at follow-up in about half of the parents, the most common feeling being worry-anxiety-fear; the ATD had a strong emotional importance for about a fifth of the parents; more than a third experienced little personal competence regarding the ATD; and about 40 % showed a need to discuss the ATD with the interviewer.

Interviewer's evaluation of parent's long-term emotional adjustment to the child's ATD

An "adequate adjustment" was judged to characterize more than half of the mothers (54 %) and two thirds of the fathers (69 %). These parents' reports indicated that their thoughts and feelings about the ATD had gone through a process of change and working-through, which had been beneficial to the parent. These parents could remember and discuss their previous and current feelings about the child's ATD without reporting or showing signs of strongly negative current feelings such as worry, anxiety, fear, anger, guilt or depression. Nothing suggested that the parent's current feelings about the ATD represented a problem for the parent or for the parent's relation to the child or to other family members. (Non-problematic negative feelings of mild to moderate degree were nevertheless observed for some of these parents.)

A "somewhat poor" emotional adjustment was judged for more than a third of the mothers (37 %) and for a fourth (25 %) of the fathers. In

these cases, the parent's feelings and thoughts associated with the ATD had not been completely worked-through and integrated within the parent during the time since the ATD had been identified. During the interview, these parents showed one or more signs of currently negative and rather strong feelings about the child's ATD; these feelings were painful or difficult for the parent to handle, which was indicated by e.g. the parent's denial, belittlement, etc. of the child's ATD or their own feelings toward it. The ATD also currently represented a problem for the parent emotionally, as seen e.g. in feelings of low self-esteem or difficulty in the parent-child relationship (e.g. over-protection) or the marital relationship (e.g. coldness or hostility).

A "very poor" emotional adjustment was judged for very few mothers (8 %) or fathers (6 %). These parents evidenced no or almost no working-through or integration of their negative feelings, and their feelings (e.g. anxiety) and thoughts about the child's ATD existed almost constantly and daily influenced the parent's own life, relationship to the child (e.g. strong insecurity and uncertainty regarding child-rearing, over-protection) and relations to other family members.

Relationship between interviewer's judgment of parent's emotional adjustment and characteristics scored by another researcher from interview transcripts

The interviewer's assessment of the parent's adaptation was based on his total impression from the home visit, while the other researcher's assessments of the four transcript-variables presented above were independently and systematically scored on the basis of that part of the interview concerned with the ATD. The comparison of these two different assessors' sco-

res (each with their particular potential advantages and limitations) gives some possibility of "cross-validating" both of them.

Analyses indicated that the interviewer's judgment of parental emotional adaptation was significantly positively related to the other researcher's independent evaluation of the above four variables: Those mothers judged by the interviewer as having "somewhat/very poor emotional adjustment" were (as compared with mothers with "adequate" adjustment) independently judged by the other scorer as more often a) having predominantly negative feelings toward the ATD at follow-up ($P < 0.0005$), b) experiencing low competence regarding the ATD ($P < 0.025$), c) assigning the ATD (very) great emotional importance at present ($P < 0.0005$) and d) showing a (very) great need to discuss the ATD with the interviewer ($P < 0.025$); they also more often felt worry-anxiety-fear, less often satisfaction, and somewhat more often uncertainty-wondering regarding the ATD at present.

Fathers judged as having somewhat/very poor emotional adaptation were independently judged by the other researcher as more often a) having predominantly negative feelings toward the ATD ($P < 0.05$) and c) assigning (very) great emotional importance to the ATD ($P < 0.005$); their increased tendency toward b) experiencing low competence regarding the ATD and d) their need to discuss the ATD with the interviewer did not reach statistical significance. These fathers also had an increased frequency of currently negative feelings such as worry-anxiety-fear, guilt and anger concerning the ATD.

Continuity of the parent's feelings about the child's ATD over time

The degree of continuity over time in the parent's basic feelings about the ATD was studied by comparing the nature of the feelings upon receiving

the first news of the child's ATD (11) (as reported at follow-up) and the nature of the feelings in evidence at follow-up 5-7 years later. For each of these occasions, each parent was categorized as having either predominantly "negative" feelings (i.e. one or more among worry-anxiety-fear, shock, guilt, anger, and depression) or predominantly "non-negative" feelings (i.e. satisfaction, calmness-composure, and/or uncertainty-wondering, but none of the negative feelings).

The relationship between the parent's feelings initially and at follow-up is shown in Table 2. A considerable degree of association was observed for mothers' feelings across these two points in time ($P = 0.012$): Three fourths (74 %) of the mothers who had had negative feelings initially still had them at follow-up, and a similar degree of stability was found for mothers' non-negative feelings (75 %).

Less association was found between fathers' initial feelings and those observed at follow-up ($P = 0.293$). Among the initially negative fathers, half (50 %) were still negative at follow-up, while 64 % of the initially non-negative fathers were still so at follow-up.

The interviews for the parents whose initial feelings had changed by the time of follow-up were studied further to determine whether the parents' reports could provide an explanation for these changes. Most (10/12) of the mothers whose initially negative feelings had become non-negative by follow-up reported that the medical system had helped them; they typically reported having received both complete information about the ATD and emotional support from the pediatrician at the later contacts. Fathers whose initially negative feelings had disappeared by follow-up reported e.g. that they had seen that the child was normal and healthy, they had received more information about ATD or they had simply become

Table 2

Relationship between parent's* initial and current feelings regarding the child's ATD[$]

Initial feelings	n (%)	Current feelings: Negative — Mothers n = 36 (67 %)	Current feelings: Negative — Fathers n = 19 (45 %)	Current feelings: Non-negative — Mothers n = 18 (33 %)	Current feelings: Non-negative — Fathers n = 23 (55 %)
Negative					
Mothers	46 (85)	34		12	
Fathers	28 (67)		14		14
Non-negative					
Mothers	8 (15)	2		6	
Fathers	14 (33)		5		9

*Five mothers and six fathers could not be classified regarding initial and/or current feelings.

[$]Association between initial and current feelings (exact P): Mothers' P = 0.012, Fathers' P = 0.293.

accustomed to the child's ATD. In contrast, both mothers and fathers who did not report initially negative feelings but had them at follow-up reported they had received more information about ATD, which had made them anxious and worried.

DISCUSSION

Significant positive relationships were observed between the interviewer's assessment of parental emotional adjustment to the child's ATD, 5-7 years after its identification, and the assessments of specific variables done independently per interview transcripts; these positive relationships represent evidence for the validity of these different evaluations.

The interviewer judged about half of the mothers and a third of the fathers as having a poor long-term emotional adjustment to the child's ATD, and for a few mothers (8 %) and fathers (6 %), the ATD was so problematic that negative effects of it were observed in their daily life. The independent assessments of the interviews indicated that 58 % of the mothers and 44 % of the fathers had predominantly negative feelings (most commonly worry-anxiety-fear and guilt) about the child's ATD at follow-up. Further signs that the ATD was currently emotionally problematic for many (20-50 %) of the parents were found in the ATD's great emotional importance for the parents, the parents' experienced lack of competence about it, and the parents' need to discuss the ATD with the interviewer. The child's ATD was usually more emotionally problematic for the mothers than for the fathers.

The present results indicate negative long-term effects on the parents, which must be considered in the cost-benefit analysis of neonatal screening for ATD (10).

A strongly positive association was found between the nature of the mothers' (but not fathers') initial feelings toward the ATD at its identification and their feelings at follow-up. Negative and non-negative initial reactions were equally stable over time.

Factors associated with changes in parents' initial negative feelings over time may suggest ways of helping parents. Almost all of the mothers and some of the fathers whose initially negative feelings had disappeared by follow-up reported that having received emotional support and satisfactory information about ATD from the physician at the medical check-ups had helped them to feel better about the child's ATD. This would suggest that giving special attention to parents of children with ATD might help them to work through the negative feelings typically evoked by the identification of the child's ATD and help them to adjust emotionally to the existence of the ATD.

In total, the present findings suggest that some of the parents of children with ATD do need assistance in coming to terms with their feelings regarding the ATD, and that the medical system may provide this help. To promote this, it would be helpful to be able to identify in advance those parents who are at especially high risk for negative reactions and for poor long-term emotional adjustment, and, further, to know what particular events and situations tend to hinder versus promote parents' emotional adjustment. Data concerning both the parents' personal characteristics and possibly relevant events and situations have been collected in the current project, and systematic analyses are being done of these variables' relationship to parental emotional adjustment. The results of these analyses will be presented subsequently.

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ARTICLE V

IDENTIFYING CHILDREN AT HIGH SOMATIC RISK (α_1ANTITRYPSIN DEFICIENCY): POSSIBLE LONG-TERM EFFECTS ON PARENTS' REPRODUCTION, MARITAL STATUS AND SOCIAL CLASS LEVEL

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ABSTRACT - Neonatal identification of children at high somatic risk due to inherited $\alpha_1$antitrypsin deficiency (ATD) has been found to elicit a negative emotional reaction in a majority of the parents, at least initially. This sub-study was conducted to determine whether the identification and follow-up of the children's ATD had negatively influenced the families' reproduction, marital status and social class level (defined by parental occupation) during the 5 years following the identification of the children's ATD. No such negative effects were found in comparing these characteristics among 61 families with a child with ATD versus 183 control families living in the same area and having a child of the same sex and age. Unexpectedly, the ATD-children's families (fathers) had a significantly higher social class level, raising the question of a possible gene advantage associated with ATD.

One in every 1500 individuals in Sweden has a high risk for chronic obstructive lung disease in adulthood and somewhat increased risk for neonatal

liver disease as a result of a deficiency of $\alpha_1$antitrypsin, a protein found in the blood (1). Antitrypsin deficiency (ATD) is inherited from both parents. While ATD cannot at present be treated or cured, a considerable prophylactic effect postponing the onset of chronic lung disease is obtained by protecting the seriously antitrypsin-deficient individual from concentrated air pollutants, especially cigarette smoke (2). From economic and somatic viewpoints, general screening for ATD is optimally conducted shortly after birth, using the same blood sample as that used for screening for PKU (3).

Nation-wide screening for ATD among all newborns (200,000) was conducted in Sweden in 1972-1974. The families of identified cases were informed of the child's ATD by a physician from the local medical facility, and the children were to attend additional physical check-ups at 6 months and 1, 2 and 4 years of age.

During the course of informing the parents and conducting the follow-up of the cases, a number of pediatricians reported clinical observations that the identification of the child's ATD appeared in some cases to have considerably negative psychological consequences for the parents and parent-child relationship. The neonatal screening for ATD was discontinued, largely due to these observations. The strong need for a systematic study of the psychological and psychosocial consequences of ATD-identification at this age level was recognized. The results of such a study would be important for subsequent decisions regarding whether or not to resume general screening for ATD, and what the most appropriate forms for such screening and contact with the cases would be. With the quickly expanding possibilities for early identification of health-risk conditions (4, 5), general interest in these topics is rapidly increasing (6-11).

The current project (12) was thus developed to investigate what effects the "ATD-matter" (i.e. a diversity of possible ATD-related influences including the identification of ATD and subsequent related events) had had on the parents and families, as studied 5-7 years after the ATD was identified. Systematic study of a representative group of parents of 61 ATD-cases found that 78 % of the mothers and 58 % of the fathers had had negative emotional reactions (especially worry-anxiety-fear) upon first learning of the child's ATD; in many cases the negative reactions were strong and long-lasting (13). A majority of the parents had reacted to the ATD as if it implied an immediate threat to the child's health. At follow-up most parents knew that the child's ATD was inherited from the parents (14).

This article presents a systematic study of the ATD-matter's possible long-term effect on the families' reproduction rates, marital status and social class level. A number of previous studies have found that these three demographic characteristics were negatively influenced by the identification of some types of serious disease in children: The birth of a handicapped or seriously ill child was associated with a) subsequently increased family planning and reduced reproduction (in various genetic disorders (15)), b) subsequently increased marital disharmony including separation and divorce (in parents of children with spina bifida (16) and prematurity requiring hospitalization (17)), and c) changes in parental occupation, sometimes representing a lowering of standards for the family (with a mentally retarded child (18, 19)). In contrast, other studies have found no reduction in reproduction among parents who are gene carriers for Huntington's chorea (20, 21) or parents of children with Down's syndrome (22, 23), and no increased frequency of divorce or separation among the

parents of children with Down's syndrome (23), cancer (24) or cystic fibrosis (25).

If the ATD-matter has negatively influenced these demographic characteristics of the families in the current study, then the ATD-group will be found to have significantly lowered reproduction, increased rates of separation and divorce and an increased frequency of decline in social class during the 5-year period following the identification of the child's ATD. These hypotheses were tested by comparing these characteristics in the ATD-group versus a specially chosen control group.

METHODOLOGY

ATD-sample

The 62 children with the homozygous PiZZ form of ATD, identified through screening 1972-74, who lived in southern Sweden (south of a line from and including Göteborg to Stockholm), whose families were Swedish-speaking and who had been contacted in association with the identification of the child's ATD, and who had not had liver disease, congenital malformations or chronic somatic diseases, were selected for study; the 61 families that agreed to participate constituted the current sample (12).

The children's age at follow-up averaged 5.8 years (range from 4.5 to 7.1), and was 5 years or older in 90 % of the sample. As 95 % of the families were known to have been informed of the child's ATD by the time the child was 6 months old (13), the observation period for effects on the demographic characteristics, after the parents were first contacted about the ATD, was thus generally 5 years or longer.

Partially-matched control (PMC) sample

The goal in choosing this control sample was to provide a reliable refe-

rence group against which to evaluate the ATD-group's demographic characteristics that were hypothesized to have been influenced by the ATD-matter. The control group would optimally control for the possible effects of basic demographic characteristics not of interest in this context (i.e. Swedish-speaking families living in same geographical area, giving birth to a child of the same sex at about the same time), while allowing the demographic characteristics of interest (reproduction, marital status, social class level) to vary as freely as possible within such constraints. The solution was to only partially match a large control group.

For each ATD-case, three PMC-cases were selected as a) the children of the same sex from Swedish-speaking families, b) born nearest in time to the ATD-case, c) registered at the same well-baby clinic and d) not suffering from ATD, a chronic somatic disease or a serious congenital malformation (in order to exclude conditions with possible psychosocial consequences similar to those hypothesized for ATD). Eleven (6 %) PMC-cases originally selected were excluded due to serious allergy-bronchial asthma, congenital malformations, bilateral ureteral reflux with repeated urinary infections, epilepsy, mental retardation, bilateral hearing defect and diabetes. These 11 cases were replaced by new PMC-cases without such somatic conditions.

The final PMC-sample (n = 183) was almost identical to the ATD-sample on the children's mean age (5.9 $\pm$ 0.6 vs. 5.8 $\pm$ 0.6), median age (5.8 vs. 5.8) and age range (4 yr. 7 mo. - 7 yr. 2 mo. vs. 4 yr. 6 mo. - 7 yr. 1 mo.) at follow-up. The observation periods for the ATD- and PMC-groups were thus equally long.

Demographic variables

The data used for comparison of the ATD- and PMC-groups were obtained

solely from the well-baby clinic and population register systems, in a comparable manner for the two groups. All data were scored by the same person.

Reproduction was expressed in terms of number of children born live into the family since the birth of the "index" child (i.e. the AID- or PMC-child chosen for study). The current number of children represented the biological offspring of either parent living in the home at follow-up.

The parents' formal marital status was classified as unmarried, married, divorced or widowed, and the parents were also characterized as cohabiting or not. Divorces occurring since the birth of the index child were counted among biological parents who were married to each other at or since the index child's birth. Separations since the birth were counted among those biological parents who were not married but were known to be cohabiting at or since the birth. (For two PMC-cases, it was impossible to determine from the information available whether the non-married parents had cohabited.)

Family social class was expressed in the usual Swedish manner in terms of three classes (26), defined by the highest among the parental occupations. The upper social class represented professionals and higher level businessmen, the middle class white collar workers and lower level businessmen, and the lower class semi- and unskilled workers. Study of social class level and changes over time was limited to families/parents with occupational data at both the child's birth and time of follow-up. (Information on the parents' occupations at the time of the birth was not available from the current sources for 13 AID- and 37 PMC-fathers, 6 AID- and 29 PMC-mothers, and in total 4 AID- and 20 PMC-families.)

Statistical analysis. The relation between AID- vs. PMC-status and each

of the demographic variables was tested by Chi-square, with Yates' correction in 2 X 2 tables, and Fisher exact probability (27). The cut-off for statistical significance was $P < 0.05$. The hypotheses indicated 1-tailed analyses.

RESULTS

Hypothesis 1. Reproduction following the birth of the index child is lower in the ATD- than the PMC-group.

At the birth of the index child, the ATD- and PMC-groups were highly similar to one another on mean number of children (1.7 vs. 1.8) and on proportion of families with one, two and three or more children. During the follow-up period, 39 % of the ATD-families and 33 % of the PMC-families had additional children. The mean number of additional children per reproducing family was identical in the two groups (Table 1). The ATD- and PMC-groups were also highly similar to one another on mean number of children in the home and on the proportion of families with one, two and three or more children at follow-up (Table 1). No support was obtained for Hypothesis 1.

Hypothesis 2. The frequency of divorce and separation during the follow-up period is higher in the ATD- than the PMC-group.

The proportion of parents who were married at or since the time of the child's birth was almost identical in the ATD- (84 %) and PMC- (86 %) groups, as was the proportion cohabiting (Table 2). The proportion of married couples who had divorced since the birth was only slightly and not significantly higher in the ATD- (16 %) than the PMC- (9 %) group; the rate of separation among unmarried cohabiting couples was quite similar in the ATD- (25 %) and PMC (32 %) groups (Table 2). The total rate of divor-

Table 1

Reproduction after birth of index child and number of children in home at follow-up

Variable		ATD-group (n = 61) n (%)	PMC-group (n = 183) n (%)
Had any additional children*			
	No	37 (61)	122 (67)
	Yes	24 (39)	61 (33)
Total number of additional children		27	66
Mean number of additional children per family having additional children		1.1	1.1
Number of children in home at follow-up**			
	One	11 (18)	28 (15)
	Two	34 (56)	111 (61)
	Three	14 (23)	34 (19)
	Four	2 (3)	8 (4)
	Five	0	2 (1)
Mean number of children in home at follow-up		2.1	2.2

* ATD vs. PMC (no/yes): $X^2 = 0.49$, 1 df, n.s.

** ATD vs. PMC (1/2/3-5): $X^2 = 0.49$, 2 df, n.s.

Table 2

Divorce and separation of biological parents after birth of index child, and current parents' marital/cohabitation status

Variable		ATD-group n (%)	PMC-group n (%)
Married at or since index child's birth		51 (84)	158 (86)
Divorced since index child's birth*		8 (16)	14 (9)
Not married but cohabiting at or since index child's birth		8 (13)	19 (10)
Separated since index child's birth**		2 (25)	6 (32)
Divorced (among married) and separated (among cohabiting), as above§		10 (17)	20 (11)
Not married or known to be cohabiting at or since index child's birth		2 (3)	6 (3)
Formal marital status at follow-up‡			
	Unmarried	9 (15)	19 (10)
	Married	45 (74)	148 (81)
	Divorced	7 (11)	15 (8)
	Widowed	0	1 (1)
Cohabiting at follow-up+		51 (84)	164 (90)

* ATD vs. PMC (divorced yes/no): $X^2 = 1.25$, 1 df, n.s.

** ATD vs. PMC (separated yes/no): Fisher exact $P = 0.78$, n.s.

§ ATD vs. PMC (divorced-separated yes/no): $X^2 = 0.81$, 1 df, n.s.

‡ ATD vs. PMC (unmarried/married/divorced-widowed): $X^2 = 1.42$, 2 df, n.s.
(married/other): $X^2 = 1.00$, 1 df, n.s.

\+ ATD vs. PMC (cohabiting yes/no): $X^2 = 1.06$, 1 df, n.s.

ces and separations was thus not significantly different in the ATD (17 %) vs. PMC- (11 %) groups. Further, no significant difference was found between the two groups on the current parents' formal marital status or cohabitation status at follow-up (Table 2). No support was found for Hypothesis 2.

Hypothesis 3. Decline in social class level since the birth of the index child is more common among ATD- than PMC-families.

No significant difference was found between ATD- and PMC-groups on changes in class level since the birth. The ATD-group was almost identical to the PMC-group on the proportion of families that had declined (5 % vs. 4 % respectively), while somewhat more ATD- (18 %) than PMC- (10 %) families had risen in social class categorization since the birth of the child (Table 3). Change rates were also similar in the two groups when calculated for the mothers and the fathers separately. All mobile cases changed one step only. No support was obtained for Hypothesis 3.

At the time of follow-up, the ATD-group was significantly higher in social class level than was the PMC-group, the ATD-families having an especially high rate of cases at the upper class level (25 % ATD vs. 10 % PMC) (Table 3). This higher family level was due to the significantly higher level of the ATD-fathers' occupations; the mothers in the ATD-group were very similar to the mothers in the PMC-group on occupational level at follow-up. The higher level of the ATD-group families/fathers tended to exist at the time of the child's birth, although the difference between ATD- and PMC-families/fathers was not statistically significant at that time.

DISCUSSION

Table 3

Changes in social class level since birth of index child and social class distributions at follow-up and at child's birth

Variable	Families		Fathers		Mothers	
	ATD n (%)	PMC n (%)	ATD n (%)	PMC n (%)	ATD n (%)	PMC n (%)
Changes since birth						
Unchanged	44 (77)	139 (85)	33 (80)	118 (91)	49 (91)	140 (91)
Declined	3 (5)	7 (4)	4 (10)	3 (2)	0	2 (1)
Risen	10 (18)	17 (10)	4 (10)	9 (7)	5 (9)	12 (8)
	(X^2 = 2.16, 2 df, n.s.)					
Level at follow-up						
Upper class	14 (25)	17 (10)	12 (24)	15 (10)	4 (7)	8 (5)
Middle class	35 (61)	108 (66)	19 (39)	61 (42)	38 (68)	102 (63)
Lower class	8 (14)	38 (23)	18 (37)	68 (47)	14 (25)	53 (33)
	(X^2 = 7.88, 2 df, $P < 0.02$)		(X^2 = 6.19, 2 df, $P < 0.05$)		(X^2 = 1.33, 2 df, n.s.)	
Level at child's birth						
Upper class	11 (19)	21 (13)	10 (21)	17 (12)	3 (5)	7 (5)
Middle class	34 (60)	90 (55)	19 (40)	56 (38)	34 (62)	89 (58)
Lower class	12 (21)	52 (32)	19 (40)	73 (50)	18 (33)	58 (38)
	(X^2 = 3.05, 2 df, n.s.)		(X^2 = 3.03, 2 df, n.s.)		(X^2 = 0.45, 2 df, n.s.)	

As compared with 183 control families who were living in the same geographical area and who had had a child of the same sex at the same time, the 61 families with a child with ATD evidenced no reduction in reproduction, no increase in divorce or separation and no increased frequency of decline in social class level during the 5 or more years since the birth of the child. Thus, no evidence was found in the current analyses to support the hypotheses that the identification and somatic follow-up of the child's ATD had negatively influenced the families' reproduction, marital status or social class characteristics.

Increased reproduction/fertility has previously been coupled with the ATD-gene (28, 29), and was thought to be due to chemically facilitated conception. While no increased fertility was evidenced in the current ATD-sample, such a possible genetic effect on fertility is difficult to study at present in Sweden, because of the widespread usage of effective contraceptives.

These ATD-parents' attitudes toward their spouses and toward having additional children have also been studied separately through parental interviews at follow-up; the ATD-parents' attitudes were found to be quite similar to those of another, more extensively demographically-matched control sample (30, 31). The results obtained from these two, independent sources of information thus tend to corroborate each other in indicating that the ATD-matter had no notable negative long-term effect on the marital relationship or parental attitude toward further reproduction. This lack of negative effect is relevant to subsequent cost-benefit analysis of the question of whether to screen for ATD early in life; the current findings suggest no "costs" regarding these demographic characteristics.

The initial characteristics of the ATD- vs. PMC-groups at the birth of

the index child would not appear to have unduly influenced the current results. The two groups were highly comparable on initial marital and cohabitation status as well as initial family size. The somewhat higher social class level observed in the ATD-group at the birth of the child could have represented, if anything, a bias in favor of the hypothesis, as the higher level of the ATD-group might have provided opportunity for more decline in the ATD-group and for more upward mobility in the controls; no such results were observed.

The significantly higher social class level observed for the ATD-families was an unexpected finding. This higher level is not explainable in terms of any known methodological bias in collecting or scoring the social class data. More complete information about occupations, obtained through interviews with the ATD-parents in the home at follow-up, produced an almost identical total social class profile for the ATD-families (12). Differential parental age cannot account for the higher social class level in the ATD-group, as no significant difference in parental age was found for the total ATD- vs. PMC-group or for those families in the upper class (Table 4). If anything, the ATD-fathers tended to be younger than PMC-fathers, which would suggest that their higher class level may have been attained at a relatively young age.

The possibility that the ATD-group's higher social class level was at least partially a psychological consequence of the ATD-matter cannot be excluded, but the tendency toward an initially higher level at the time of the child's birth (and before the parents knew of the child's ATD and their own gene-carrier status) was larger than the change since the child's birth, which might suggest the importance of factors other than psychological reactions.

Table 4

Parental age at follow-up, in total sample and in families classified as upper class (I)

Parental group	Age group (years)					
	20-24	25-29	30-34	35-39	40-44	45 +
	n (%)	n (%)	n (%)	n (%)	n (%)	n (%)
All ATD-mothers*	2 (3)	16 (27)	27 (45)	10 (17)	4 (7)	1 (2)
All PMC-mothers	7 (4)	49 (27)	77 (42)	36 (20)	11 (6)	2 (1)
All ATD-fathers**	1 (2)	4 (8)	26 (50)	14 (27)	6 (12)	1 (2)
All PMC-fathers	5 (3)	24 (15)	56 (34)	51 (31)	20 (12)	7 (4)
Class I ATD-mothers$		2 (15)	7 (54)	3 (23)		1 (8)
Class I PMC-mothers		3 (16)	8 (42)	7 (37)	1 (5)	
Class I ATD-fathers+		1 (8)	9 (69)	2 (15)	1 (8)	
Class I PMC-fathers			9 (50)	7 (39)	2 (11)	

* ATD vs. PMC (20-29/30-34/35-39/40+): $X^2 = 0.40$, 3 df, n.s.

** ATD vs. PMC (20-29/30-34/35-39/40+): $X^2 = 4.68$, 3 df, n.s.

$ ATD vs. PMC ($\leq$ 34/$\geq$ 35): Exact P = 0.79, n.s.

+ ATD vs. PMC ($\leq$ 34/$\geq$ 35): $X^2 = 1.31$, 1 df, n.s.

Question may be raised as to whether the factors lying behind the higher social class level of the ATD-fathers might possibly represent a selective gene advantage of the heterozygote over the normal homozygote. Population geneticists have classified ATD as representing one example of balanced polymorphism (32). In such a case, some special characteristic(s) providing increased viability would be found in the heterozygote, and the biological advantage would (from a genetic perspective) compensate for the predisposition to disease in homozygous cases. For example, in sickle cell anemia the increased death rate in the homozygote is balanced by the increased resistance to malaria found in the heterozygote (32). Similarly, the significantly increased intelligence found in the siblings of children with PKU may indicate the possibility of superior qualities associated with carrier status (33-35).

Before any conclusions are drawn about a possible gene advantage associated with higher social class level (occupational level) in the ATD-group, the current findings need to be replicated on independent samples. One possible sample would be the remaining ATD-cases who were identified by the 1972-74 screening and who lived outside the current sampling area. If the current results can be independently replicated, further consideration also needs to be given to the questions of what aspect(s) of higher social class level (defined per parental occupational classification) might reflect a genetic survival factor, and why such an advantage is observed only among fathers (when both fathers and mothers are gene-carriers).

Another interesting incidental finding was the higher proportion of separations among nonmarried cohabiting pairs (25-32 %) than divorces among married pairs (9-16 %). Question may be raised as to whether the

formality of marriage provides more stability over time and/or whether persons who do not formalize their relationship through marriage are themselves different or have a less solid relationship.

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ARTICLE VI

IDENTIFYING CHILDREN AT HIGH SOMATIC RISK: POSSIBLE LONG-TERM EFFECTS ON THE PARENTS' ATTITUDES TOWARD THEMSELVES AS PARENTS AND TOWARD HAVING MORE CHILDREN

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ABSTRACT - Neonatal identification of children at high somatic risk due to inherited $\alpha_1$antitrypsin deficiency (ATD) was hypothesized to have had negative long-term effects on the parents' attitudes toward themselves as parents and toward having more children. The parents of 61 children with ATD were followed-up and studied by interviews about 5 years after the child's ATD was identified, and compared with demographically similar parents of 61 control children without ATD. No support was found for the hypothesized negative effects on these parental attitudes.

Nation-wide neonatal screening for $\alpha_1$antitrypsin deficiency (ATD) was conducted in Sweden from 1972-1974, but discontinued thereafter due to clinical observations that the early identification of ATD appeared in some cases to have notably negative psychological effects on the parents and parent-child relationship. A cost-benefit analysis of the screening was requested by medical authorities. A systematic study was begun to

investigate the effects of the ATD-matter (i.e. the identification and somatic follow-up of the ATD) on the parents, families and children, as studied 5-7 years after the children's ATD was identified (1).

Among the possible long-term effects, it was hypothesized that the ATD-matter had negatively influenced the parents' attitudes toward themselves as parents, especially in relation to this child. For many parents, a sick or deviant child represents a strong threat to their self-concept, especially when the illness or deviation is identified in the newborn period (2, 3). Parental feelings of inadequacy, inferiority, guilt or shame related to illness or deviance in the child can derive from several different factors: The child may represent a personal failure and signify their inability to create or produce a healthy child (3-5). The presence of genetically-based disorders in the child may give the parents feelings of inferiority because they are the carriers of "bad genes" (3); the parents may feel they have "caused" the child's illness (3, 6, 7) and are inferior parents for having "damaged" the child. Further, the parents may feel shameful and inferior for having the negative feelings that are often aroused by an ill or deviant child (7).

It was also hypothesized that the ATD-matter had negatively influenced the parents' current attitudes toward having more children. Such an effect on parents' attitudes may also have led to increased intra-family disagreement about whether to have more children. Previous studies have shown that parents' attitudes toward having more children are generally put to a hard test after having a seriously ill or handicapped child, and the question of whether to have more children often represents a conflict both within the parent and between the parents (3). Increased family planning and reduced reproduction have been found among the parents of children

with some (8) but not all (6, 9-11) genetic disorders.

These hypothesized long-term effects of the ATD-matter were tested by comparing these characteristics in parents of children with ATD vs. demographically similar control parents of children without ATD, studied by interview at follow-up. The operational hypotheses were that ATD-group parents (as compared with controls) are more negative toward themselves as parents, more often do not plan to have more children, and more often disagree within the family regarding this.

SAMPLES AND METHODS

The samples and methodology are described in greater detail elsewhere (1, 12, 13).

The ATD-sample consisted of families of 61 children with the homozygous Pi*ZZ* form of ATD, identified through screening in 1972-74, living in southern Sweden, and free from liver disease, congenital malformations and chronic somatic diseases. Their mean age at study was 5.8 years. Interview data were obtained with 59 mothers and 49 fathers.

The matched control (MC) sample consisted of 61 families with a child free from ATD, serious congenital malformations and chronic somatic diseases, and individually matched with the ATD-cases on well-baby clinic, age, sex, social class, number of parents and siblings, sibling order and type of housing (1). Interviews were obtained with 61 MC-mothers and 45 fathers.

ATD- and MC-groups were similar to one another on maternal age and number of children in the home at follow-up (mean = 2.2 and 2.1 children in ATD- and MC-groups, respectively) (1, 12). A vast majority of mothers in both groups were still in the fertile age range at interview; 92 % of

the ATD-mothers and 97 % of the MC-mothers were under 40 years of age and all mothers except one were younger than 45.

Each parent was interviewed separately in the home by a physician (T.T.). The parents were asked i.a. how they experienced being a parent to the child (and its siblings) and how they thought others (friends, their own parents, the child) viewed them as parents. (It was assumed that the parents would tend to project their own attitudes into the attitudes they reported for other persons.) Clinical summary judgments of the parents' attitudes toward themselves were also made on the basis of the total interview contents.

Transcripts of the interviews were edited to remove (temporarily) information that could bias scoring by another researcher (E.A.J.) who was to be blind as to the parent's ATD- vs. MC-status (see 12, 13). The transcripts were then scored per a detailed scoring system (12). Interscorer agreement was tested.

The following six variables (and their interscorer agreements) were used to test the hypothesized effect on parents' attitudes toward themselves; each variable was scored on a 5-point scale (from "very positive" to "very negative"): a) Scorer's judgment of the parent's attitude toward him-/herself as a parent (r = + 0.79); b) Parent's view of "how the child would describe the parent, e.g. to a friend" (+ 0.80); c) Parent's view of how his/her own parents view him/her as a parent (+ 0.94); d) Parent's view of how friends and acquaintances view him/her as a parent (+ 0.86); e) Parent's experience of being a parent to the child (+ 0.83); f) Parent's experience of being a parent to the child, as compared with to its siblings (+ 0.80). None of the scores for these six variables were influenced by the removal of potentially biassing information from the

ATD-parents' transcripts.

Further, to test the hypothesized effect on parental plans for having more children, both ATD- and MC-mothers and fathers were asked, independently within each family, whether they planned to have more children in the future (scored as "yes/uncertain/no"; interscorer agreement = 95 %). One ATD-parent's score for this item was temporarily influenced by removal of information that indicated that this was an ATD-group parent, but this score was later adjusted after the blind scoring was completed for the entire sample.

Statistical analyses of the data were performed by Wilcoxon matched-pairs signed-ranks test and Chi-square with Yates' correction (13). One-tailed analyses were indicated by the hypotheses.

RESULTS

Hypothesis 1. As compared with controls, ATD-parents are more negative toward themselves as parents, especially in relation to the child.

No statistically significant differences between ATD- and MC-mothers or fathers were found on any of the six variables representing parental attitudes and experiences (Table 1). On five of the six variables the fathers' results showed (n.s.) trends against the hypothesis, the ATD-fathers tending to have a more positive attitude toward themselves as parents. Hypothesis 1 was not supported.

Hypothesis 2. As compared with controls, a) the ATD-parents more often do not plan to have more children, and b) the ATD-parents are more often in disagreement within the family regarding whether to have more children.

No statistically significant difference was found between ATD- vs. MC-mothers or fathers regarding their current plans to have more children (Table

Table 1

ATD and control parents' attitudes toward themselves and experiences of being a parent to the child

Variable	n	Category n (%)					Pair-wise ATD-control comparison			
							Pairs n	ATD worse than control n (%)	Control worse than ATD n (%)	Wilcoxon (1-tailed)
How child would describe parent		Very positive	Positive	Mixed	Negative	Very negative				
ATD-mothers	57	2 (4)	11 (19)	29 (51)	12 (21)	3 (5)	56	20 (36)	13 (23)	n.s.
Control mothers	59	1 (2)	14 (24)	36 (61)	7 (12)	1 (2)				
ATD-fathers	45	6 (13)	15 (33)	22 (49)	2 (4)	0	38	10 (26)	13 (34)	n.s.
Control fathers	45	1 (2)	15 (33)	26 (58)	3 (7)	0				
Own parents' views of parent as parent										
ATD-mothers	54	1 (2)	20 (37)	14 (26)	17 (31)	2 (4)	51	17 (33)	15 (29)	n.s.
Control mothers	57	1 (2)	19 (33)	16 (28)	21 (37)	0				
ATD-fathers	41	0	25 (61)	10 (24)	6 (15)	0	30	6 (20)	12 (40)	n.s.
Control fathers	41	1 (2)	17 (41)	16 (39)	7 (17)	0				
Friends' views of parent as parent										
ATD-mothers	56	3 (5)	8 (14)	30 (54)	14 (25)	1 (2)	54	21 (39)	16 (30)	n.s.
Control mothers	58	1 (2)	16 (28)	26 (45)	15 (26)	0				
ATD-fathers	45	0	11 (24)	26 (58)	8 (18)	0	37	8 (22)	16 (43)	n.s.
Control fathers	43	0	7 (16)	24 (56)	12 (28)	0				

Table 1, continued

ATD and control parents' attitudes toward themselves and experiences of being a parent to the child

Variable	n	Category n (%)					Pair-wise ATD-control comparison			
							Pairs n	ATD worse than control n (%)	Control worse than ATD n (%)	Wilcoxon (1-tailed)
Parent's attitude toward self as parent		Very positive	Positive	Mixed	Negative	Very negative				
ATD-mothers	59	0	20 (34)	27 (46)	12 (20)	0	59	15 (25)	22 (37)	n.s.
Control mothers	61	0	11 (18)	39 (64)	11 (18)	0				
ATD-fathers	48	0	20 (42)	26 (54)	2 (4)	0	40	8 (20)	12 (30)	n.s.
Control fathers	45	0	16 (36)	24 (53)	5 (11)	0				
Experience of being:										
A parent to the child										
ATD-mothers	59	21 (36)	25 (42)	4 (7)	7 (12)	2 (3)	59	18 (31)	24 (41)	n.s.
Control mothers	61	16 (26)	26 (43)	15 (25)	2 (3)	2 (3)				
ATD-fathers	48	16 (33)	26 (54)	6 (13)	0	0	40	14 (35)	11 (28)	n.s.
Control fathers	45	14 (31)	26 (58)	5 (11)	0	0				
A parent to the child, compared with sibs		Much easier	Easier	Same	Harder	Much harder				
ATD-mothers	43	3 (7)	10 (23)	20 (47)	9 (21)	1 (2)	40	15 (38)	14 (35)	n.s.
Control mothers	50	4 (8)	16 (32)	16 (32)	11 (22)	3 (6)				
ATD-fathers	40	0	10 (25)	27 (68)	3 (8)	0	33	8 (24)	11 (33)	n.s.
Control fathers	40	2 (5)	7 (18)	22 (55)	8 (20)	1 (3)				

2). Both ATD-mothers and fathers showed nonsignificant trends against the hypothesis, in that somewhat fewer ATD-parents than MC-parents clearly did not plan more children. Furthermore, no statistically significant difference was found between ATD- and MC-families on parental (dis)agreement within the family concerning whether to have more children. No support was found for either aspect of Hypothesis 2.

DISCUSSION

No evidence was found to support the hypotheses that the ATD-matter had had negative long-term effects on the parents' views of themselves as parents or on their attitudes toward having more children, studied at the time of follow-up about 5 years after the children's ATD was identified. These latter results regarding attitudes toward having more children coincide with those obtained in a separate part of the interview, scored independently at a later time; in the other interview part, few ATD-parents (8 % mothers, 2 % fathers) mentioned having been concerned about having more children because of the risk for ATD, and few (2 % mothers, 2 % fathers) mentioned having abstained from having more children because of this (14). Furthermore, the ATD-parents (as compared with a partially matched control group) did not have reduced reproduction during the 5-year period following the identification of the child's ATD (15).

The lack of a negative effect on parental attitudes toward themselves as parents and toward having more children was consistent and notable. In fact, the ATD-fathers showed a consistent tendency to be more "positive" than their controls, i.e. they tended to view themselves more positively as parents and to be less negative toward having additional children. As isolated, nonsignificant findings opposite to the hypotheses, these trends

Table 2

ATD and control parents' current attitudes toward having more children

Variable	n	Category n (%)			ATD-control comparison Chi-square
Plans to have more children		No	Uncertain	Yes	(No vs. Uncertain/Yes)
ATD-mothers	58	40 (69)	14 (24)	4 (7)	X^2 = 2.46, 1 df, n.s.
Control mothers	59	49 (83)	8 (14)	2 (3)	
ATD-fathers	47	33 (70)	10 (21)	4 (9)	X^2 = 1.22, 1 df, n.s.
Control fathers	45	37 (82)	7 (16)	1 (2)	
Within-family parental agreement		Disagree	Agree		
ATD-pairs	45	10 (22)	35 (78)		X^2 = 0.53, 1 df, n.s.
Control pairs	43	6 (14)	37 (86)		

appear unremarkable. Yet, these fathers have also been found to express somewhat (n.s.) greater enjoyment in their work (13), to work somewhat more than controls (unpublished data), and to be unusually high in social class (occupational) level (15). Question may be raised as to whether these "positive" characteristics of ATD-fathers represent a compensation reaction to the possible threat associated with the child's ATD. Other authors (16, 17) have discussed the possibility that the identification of disease in a child can represent a turning point for the parent, resulting in the development of a positive attitude toward the parenting role and a feeling of responsibility toward the child and interest in participating in its care. Before drawing such a conclusion, the trends observed here for the ATD-fathers need to be integrated with the remaining data for the project, to determine whether "general positiveness" is characteristic of this parent group. If so, it would be important to determine whether it represents a psychological consequence of the ATD-matter or whether it existed prior to the identification of the child's ATD.

The results of the current analyses clearly suggest that the ATD-matter did not negatively influence the parents' attitudes toward themselves as parents or their attitudes toward having more children. These findings of no negative effect ("costs") on these parental attitudes should be integrated into the total cost-benefit analysis of neonatal screening for ATD.

Acknowledgments

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ARTICLE VII

IDENTIFYING CHILDREN AT HIGH SOMATIC RISK: POSSIBLE LONG-TERM EFFECTS ON THE PARENTS' VIEWS OF THEIR OWN HEALTH AND CURRENT LIFE SITUATION

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ABSTRACT - Neonatal identification of children at high somatic risk due to inherited $\alpha_1$antitrypsin deficiency (ATD) was hypothesized to have had negative long-term effects on the parents' views of their own health and current life situation. The parents of 61 children with ATD were followed-up and studied by interviews about 5 years after the child's ATD was identified, and compared with demographically similar parents of 61 control children without ATD. No negative effect was found on mothers' or fathers' views of their current life situation or the fathers' own health. As compared with controls, ATD-group mothers reported significantly poorer mental and physical health during the past year, which was interpreted as a consequence of the stress associated with the identification of the child's ATD.

An inherited deficiency of $\alpha_1$antitrypsin (ATD), a blood protein, carries a high risk for chronic obstructive lung disease in adulthood, but the risk can be diminished considerably through avoidance of concentrated air

pollutants, especially cigarette smoke (1). Nation-wide neonatal screening for ATD was conducted in Sweden from 1972-1974, but discontinued thereafter due to clinical observations that the early identification of ATD appeared in some cases to have notably negative psychological effects on the parents and parent-child relationship. A systematic study was begun to investigate the effects of the ATD-matter (i.e. the identification and somatic follow-up of the ATD) on the parents, families and children, as studied 5-7 years after the children's ATD was identified (2).

A majority of the parents were found to have reacted negatively emotionally upon learning of the child's ATD, and in many cases these reactions were strong and long-lasting (3). Most of the parents initially conceived of ATD as representing a possibly serious imminent threat to the child's health.

One of the hypotheses concerning the long-term effects of the ATD-matter was that it had negatively influenced the parents' views of their own health and current life situation. Previous research had shown that learning that one's child is physically deviant or afflicted by a serious illness represents considerable stress for most parents; long-lasting mental problems in the form of anxiety, depression, sorrow and guilt feelings, as well as increased somatic complaints, have been observed in the parents of seriously ill children (4-10). While the problems associated with the child's disorder may represent a unifying factor around which the marital relationship may be strengthened (7, 8, 11, 12), the marital relationship is more often worsened, with increased tension, aggressiveness or coldness found between the parents (8, 11-13). Further, both the stress and practical difficulties associated with having and caring for a sick or deviant child may produce substantial changes in the parents' experience of their

life situation, negatively influencing e.g. their views of their leisure time, work, housing etc. (4, 13).

This article presents an empirical test of the hypothesis that the ATD-matter had negatively influenced the parents' views of their own health and current life situation. The hypothesis was tested by comparing these characteristics for parents of children with ATD vs. demographically similar control parents of children without ATD, studied by interview at follow-up.

SAMPLES AND METHODS

ATD-sample

The 62 children with the homozygous PiZZ form of ATD, identified through screening in 1972-74, who lived in southern Sweden, whose families were Swedish-speaking and who had been contacted in association with the identification of the child's ATD, and who had not had liver disease, congenital malformations or chronic somatic diseases, were selected for study; the 61 families that agreed to participate constituted the current sample (2). The children's age at follow-up averaged 5.8 years (range 4.5 to 7.1). Interview data were obtained for 59 (98 %) of the 60 mothers and 49 (89 %) of the 55 fathers in these families.

One of the six nonparticipating fathers did not wish to participate in the interview, and the other five were not at home at the time because of their work. The six fathers who did not participate were similar to the remaining fathers on marital status and biological parent status, but were generally higher in social class. As described by the mothers, these six fathers had reacted to the ATD-matter in a manner that was comparable to that for the fathers with interviews (14). The loss of data for these six

fathers thus does not appear to have biassed the results of the study.

Matched control sample

For each ATD-case participating in the study, one matched control (MC) was individually chosen as the child a) registered at the same well-baby clinic, b) who did not have ATD, a chronic somatic disease or serious congenital malformation, c) whose family was Swedish-speaking and d) who was born nearest in time to the ATD-case, given similarity to the ATD-case on e) sex, f) family social class, g) current number of parents in the family, h) number of siblings in the home, i) order among siblings and j) type of housing (2).

Four (7 %) MCs initially chosen were unwilling to participate, and in each case a new MC was selected and accepted participation. The pair-wise matching led to a high degree of similarity between the ATD- and MC-groups on the demographic characteristics, including the children's age (2).

All of the 61 mothers and 45 (85 %) of the 53 fathers in the final MC-sample took part in the interviews. Two of the eight nonparticipating MC-fathers did not wish to be interviewed, while the other six were not at home because of their work. No difference in social class level or any other notable characteristic was found between those fathers who did and did not participate.

Parental interviews

Each parent was interviewed separately in the home by a physician (T.T.) concerning i.a. their own physical and mental health and view of their current life situation. The questions were formulated in an identical manner for the ATD- and MC-groups. The data obtained were based partially on the parent's answers to direct questions and partially on clinical judgments summarizing the interview contents.

The interviews were tape-recorded and transcribed. The transcripts were edited by the interviewer, in order to mask the ATD- vs. MC-group status of the subject and as far as possible permit blind scoring of the data. For ATD-group parents, all interview material directly referring to the child's ATD or the ATD-matter was removed and placed in a separate transcript which was independently scored at a later time (see 15 for details). The interview material that was removed was not relevant to the scoring of the variables presented in this article. All material in the transcripts for MC-parents was retained, but these transcripts were systematically disrupted to give an appearance similar to that of the transcripts for the ATD-parents.

The edited transcripts were scored by an experienced child psychologist (E.A.J.), who was blind as to the parent's ATD- vs. MC-status as well as all other data for the subject collected by other methods (2). All variables were scored in a standardized manner, per a detailed scoring system (15). Inter-scorer agreement was tested between the scorer and another researcher (T.F.M.) on 20 randomly chosen cases. ATD- and MC-cases were scored in random order, all mothers first, followed by all fathers.

Eleven variables were used to test the present hypothesis. Nine variables were analyzed per 5-point rating scales, while the remaining two variables (c, g) were analyzed as category scales (yes/no). The variables concerning parental view of her/his mental health (with inter-scorer agreements shown in parentheses) were a) Parent had experienced mental problems during the past year (r = + 0.89 on rating scale); b) Parent's view of her/his mental health at present, as compared with 5-10 years ago (+ 0.94); c) Parent had had contact with a physician during the past year regarding her/his mental health (100 % identical score on category scale);

d) Scorer's judgment of degree to which parent experiences her/his mental condition as a problem (+ 0.92); concerning parental view of her/his physical health: e) Parent's view of her/his physical health during the past year (+ 0.81); f) Parent's view of her/his physical health at present, as compared with 5-10 years ago (+ 0.87); g) Parent had had contact with a physician during the past year regarding her/his physical health (90 %); h) Amount parent was on sick leave during the past year (+ 0.85); and concerning parental view of current life situation: i) Parental experience of enjoyment of work (includes housewives) (+ 0.79); j) Scorer's judgment of parent's attitude toward spouse (+ 0.88); k) Scorer's judgment of parent's degree of satisfaction with life in total (+ 0.77).

Statistical analysis

Comparison of ATD- vs. MC-samples on rating scale items was done by Wilcoxon matched-pairs signed-ranks test, which compares the ATD- and MC-subjects' scores within each pair, and gives greater weight to pairs with a greater intrapair difference (16). Analysis of category scale items was done by Chi-square with Yates' correction. The cut-off for statistical significance was $P \leq 0.05$. The hypothesis indicated 1-tailed tests.

RESULTS

Parents' views of their own health

Mothers: Statistically significant differences between ATD- and MC-mothers supporting the hypothesis were found on three of the eight relevant variables (Tables 1, 3, 4). In comparison with MCs, ATD-mothers reported a) having experienced significantly more mental problems during the past year, and they were judged by the scorer as d) experiencing their mental condition as more problematic during the past year. (The correla-

Table 1

ATD and control parents' views of their mental health

Variable	n	Category n (%)					Pair-wise ATD–control comparison			
							Pairs n	ATD worse than control n (%)	Control worse than ATD n (%)	Wilcoxon (1-tailed)
Experienced mental problems - past year		Never	Once or twice	Now and then	Often	Almost always				
ATD-mothers	59	4 (7)	23 (39)	19 (32)	11 (19)	2 (3)	58	28 (48)	16 (28)	P = 0.022
Control mothers	60	10 (17)	25 (42)	19 (32)	3 (5)	3 (5)				
ATD-fathers	48	11 (23)	18 (38)	14 (29)	3 (6)	2 (4)	40	15 (38)	13 (33)	n.s.
Control fathers	45	8 (18)	21 (47)	13 (29)	1 (2)	2 (4)				
Health now, as compared with earlier		Much better	Better now	Same	Worse now	Much worse				
ATD-mothers	55	5 (9)	11 (20)	21 (38)	16 (29)	2 (4)	54	14 (26)	18 (33)	n.s.
Control mothers	59	3 (5)	14 (24)	24 (41)	18 (31)	0				
ATD-fathers	44	3 (7)	12 (27)	21 (48)	8 (18)	0	37	15 (41)	15 (41)	n.s.
Control fathers	45	1 (2)	15 (33)	20 (44)	8 (18)	1 (2)				
Parent views mental health as a problem		No	A mild problem	Somewhat, sometimes	Clearly a problem	A big problem				
ATD-mothers	58	16 (28)	15 (26)	13 (22)	12 (21)	2 (3)	57	28 (49)	19 (33)	P = 0.049
Control mothers	60	21 (35)	18 (30)	12 (20)	7 (12)	2 (3)				
ATD-fathers	48	24 (50)	14 (29)	8 (17)	1 (2)	1 (2)	40	14 (35)	16 (40)	n.s.
Control fathers	45	19 (42)	17 (38)	6 (13)	1 (2)	2 (4)				

Table 2

Predominant mental complaints by mothers reporting relatively "poor" mental health during the past year*

Symptom	AID-mothers (n = 32)	Control mothers (n = 25)
General nervousness, anxiety, uneasiness, strain	16	14
Depression or depressive mood	9	7
Sleep problems	6	1
Phobic symptoms	1	0
Emotional lability	0	1
Obsessive symptoms	0	1
Hypochondriasis	0	1

*Scale points (now and then, often, almost always mental problems) in Table 1.

tion between variables a) and d) was + 0.80 for the ATD-mothers and + 0.85 for ATD-fathers.)

About one-fifth (22 %) of the ATD-mothers (vs. 10 % MC) reported "poor" or "very poor" mental health, i.e. they "often" or "almost always" had felt anxiety, depression or had trouble sleeping during the past year (Table 2); and fewer ATD- (46 %) than MC-mothers (58 %) reported they had never or very seldom experienced such mental problems. The scorer judged mental health as being "clearly a problem" or "a big problem" for 24 % of the ATD-mothers (vs. 15 % MCs) and no problem or only a mild problem for 53 % of the ATD-mothers (vs. 65 % MCs) (Table 1).

Further, the ATD-mothers reported experiencing e) significantly poorer physical health during the past year (Table 3). While only 39 % of the ATD-mothers (vs. 55 % MC) reported feeling well or very well, 22 % of the ATD-mothers (vs. 13 % controls) reported experiencing poor or very poor physical health (e.g. constant pain, periods of sick leave, chronic disease implying restriction on daily life) (Table 3).

In contrast, no significant difference or even notable trend was found for ATD- vs. MC-mothers on experienced changes in either b) their mental health or f) their physical health, as compared with 5-10 years earlier (Tables 1 & 3), or c) the frequency with which the mothers had had contact with a physician during the past year concerning their mental or physical health (Table 4).

Fathers: No statistically significant difference (or even notable trend) was found between ATD- and MC-fathers on any of the eight variables related to mental or physical health.

Parents' views of their current life situation

Table 3

ATD and control parents' views of their physical health; sick leave during past year

Variable	n	Category n (%)					Pair-wise ATD–control comparison			
							Pairs	ATD worse than control	Control worse than ATD	Wilcoxon (1-tailed)
							n	n (%)	n (%)	
View of physical health		Very good	Good	So-so	Poor	Very poor				
ATD–mothers	59	7 (12)	16 (27)	23 (39)	13 (22)	0	58	26 (45)	17 (29)	P = 0.047
Control mothers	60	10 (17)	23 (38)	19 (32)	7 (12)	1 (2)				
ATD–fathers	47	5 (11)	11 (23)	19 (40)	11 (23)	1 (2)	39	9 (23)	15 (38)	n.s.
Control fathers	45	2 (4)	12 (27)	16 (36)	13 (29)	2 (4)				
Health now, as compared with earlier		Much better	Better now	Same	Worse now	Much worse				
ATD–mothers	58	1 (2)	16 (28)	26 (45)	15 (26)	0	57	19 (33)	16 (28)	n.s.
Control mothers	60	1 (2)	15 (25)	33 (55)	10 (17)	1 (2)				
ATD–fathers	47	0	11 (23)	18 (38)	18 (38)	0	39	12 (31)	14 (36)	n.s.
Control fathers	45	1 (2)	11 (24)	14 (31)	18 (40)	1 (2)				
Extent of sick leave		Never	A few days	One week	Several weeks	$\geq$ 1 mo., pension				
ATD–mothers	56	36 (64)	12 (21)	6 (11)	2 (4)	0	54	13 (24)	6 (11)	n.s.
Control mothers	59	45 (76)	8 (14)	2 (3)	4 (7)	0				
ATD–fathers	44	23 (52)	11 (25)	7 (16)	2 (5)	1 (2)	34	12 (35)	13 (38)	n.s.
Control fathers	43	18 (42)	13 (30)	2 (5)	6 (14)	4 (9)				

Table 4

ATD and control parents' contact with physicians during the past year

Variable	n	Category n (%)		ATD-control comparison Chi-square
Had contact about mental health		No	Yes	
ATD-mothers	58	54 (93)	4 (7)	X^2 = 0.08, 1 df, n.s.
Control mothers	60	54 (90)	6 (10)	
ATD-fathers	48	45 (94)	3 (6)	X^2 = 0.00, 1 df, n.s.
Control fathers	45	42 (93)	3 (7)	
Had contact about physical health				
ATD-mothers	59	30 (51)	29 (49)	X^2 = 1.08, 1 df, n.s.
Control mothers	58	36 (62)	22 (38)	
ATD-fathers	46	25 (54)	21 (46)	X^2 = 0.17, 1 df, n.s.
Control fathers	44	21 (48)	23 (52)	

No statistically significant difference was found between ATD- vs. MC-mothers or fathers on satisfaction with their work, their attitude toward their spouse or their satisfaction with their current life situation in total (Table 5). ATD-group fathers showed a tendency to enjoy their work more than did MC-fathers (i.e. a trend opposite to that hypothesized).

DISCUSSION

As compared with demographically similar control (MC) parents, parents of 5-7-year old children with ATD did not evidence significantly more negative attitudes toward their spouse or less satisfaction with their work and their life situation in total. Further, ATD-group fathers did not report worse mental or physical health than did MC-fathers. In contrast, ATD-mothers (compared with MC-mothers) reported poorer mental and physical health during the previous year. The signed-ranks tests indicated a statistically significant within-pair health disadvantage for the ATD-mothers. This difference in reported maternal health in the ATD- vs. MC-samples was observed <u>across the entire scoring scale</u>, i.e. both a decreased frequency of good health and an increased frequency of poor health were reported by the ATD-mothers. Nevertheless, they did not report having contacted physicians for their health or being on sick leave significantly more often during the past year than did controls.

One approximate measure of the magnitude of the reported health disadvantage is the frequency with which ATD-mothers reported worse health than did their matched controls (and vice versa). On both physical and mental health, about half of the ATD-mothers reported feeling worse than did their matched controls (45 % worse physical health, 48 % worse mental health), while about one-third of the ATD-mothers reported feeling better

Table 5

ATD and control parents' views of their current life situation

Variable	n	Category n (%)					Pair-wise ATD-control comparison			
							Pairs n	ATD worse than control n (%)	Control worse than ATD n (%)	Wilcoxon (1-tailed)
Enjoyment of work		Very positive	Positive	Mixed	Negative	Very negative				
ATD-mothers	58	27 (47)	17 (29)	8 (14)	4 (7)	2 (3)	58	17 (29)	23 (40)	n.s.
Control mothers	61	18 (30)	29 (48)	8 (13)	5 (8)	1 (2)				
ATD-fathers	46	11 (24)	25 (54)	9 (20)	1 (2)	0	38	10 (26)	19 (50)	n.s.
Control fathers	44	8 (18)	19 (43)	12 (27)	4 (9)	1 (2)				
Attitude toward spouse										
ATD-mothers	33	0	15 (45)	13 (39)	4 (12)	1 (3)	19	4 (21)	5 (26)	n.s.
Control mothers	32	2 (6)	13 (41)	11 (34)	6 (19)	0				
ATD-fathers	37	2 (5)	17 (46)	15 (41)	3 (8)	0	21	5 (24)	7 (33)	n.s.
Control fathers	32	2 (6)	13 (41)	15 (47)	2 (6)	0				
Satisfaction with life in total		Dominates picture	Strong	One or more signs	Suggestion	No sign				
ATD-mothers	59	0	12 (20)	28 (47)	16 (27)	3 (5)	59	15 (25)	19 (32)	n.s.
Control mothers	61	0	5 (8)	34 (56)	19 (31)	3 (5)				
ATD-fathers	47	0	10 (21)	27 (57)	10 (21)	0	40	8 (20)	16 (40)	n.s.
Control fathers	45	0	7 (16)	25 (56)	13 (29)	0				

than did their controls on physical health (29 % better) and on mental health (28 % better).

Relatively little overlap was found between reported poor mental health and poor physical health in the ATD-mothers. Among the 21 ATD-mothers who had reported what was judged to represent "poor" or "very poor" mental and/or physical health, only five (24 %) had both poor mental and poor physical health, while eight reported poor physical health only and eight reported poor mental health only. (A similar proportion of overlap (17 %) was found among MC-mothers reporting poor mental and/or physical health.) In total, poor or very poor physical and/or mental health was reported by 36 % of the ATD-mothers as compared with 20 % of the controls.

The reported poorer mental and physical health for ATD-mothers might possibly be the result of several different factors: a) The ATD-gene. That the mothers are heterozygous carriers of the ATD-gene might in theory imply poorer physical health, with the possibility of secondary consequences on mental health. To test the relevance of this interpretation, the specific physical problems cited by the ATD-mothers reporting "so-so", "poor" or "very poor" physical health were outlined from the interviews. Among the 36 ATD-mothers reporting relatively poor physical health, 29 (81 %) reported headaches or problems with their back, stomach or joints as their major physical problem (Table 6); these disturbances are not known to be somatic consequences of ATD, and the interviewer (a psychiatrist) judged most of these physical complaints as being of predominantly psychosomatic nature. Repeated respiratory infections, which may be an early sign of chronic obstructive lung disease due to ATD, were reported as the major health problem by only two (6 %) of these 36 ATD-mothers. The somatic-complaint profile for ATD-mothers reporting poor physical health

Table 6

Predominant physical complaints by mothers reporting relatively poor physical health during the past year*

Symptom area	ATD-mothers (n = 36)	Control mothers (n = 27)
Repeated/chronic headaches	9	8
Repeated/chronic back problems	8	5
Repeated/chronic stomach/digestive problems (gastritis, intestinal pain, "nervous stomach")	7	4
Repeated/chronic pain in joints (knee, hip, neck, wrist)	5	2
Repeated upper respiratory infections	2	2
Repeated/chronic pain and cramps in legs	1	1
Repeated urinary tract infections	1	1
Psoriasis	1	-
Periodontoclasia (loosening of the teeth)	1	-
Chronic fatigue	1	-
Chronic hepatitis (inflammation of the liver)	-	1
Allergy	-	1
Rheumatism	-	1
Gynecological disorder	-	1

* Scale points (so-so, poor, very poor health) in Table 3.

was quite like that for the MC-mothers reporting poor physical health (Table 6). Furthermore, the ATD-fathers, who are also heterozygous ATD-gene carriers, did not report increased physical or mental ill health (compared with their controls). In total, no evidence was obtained to support the interpretation that the poorer mental and physical health reported by the ATD-mothers was a physical consequence of ATD-gene carrier status.

b) Genetic linkage with personality. Robinson (17, p. 34) has suggested the possibility that the genotype for ATD might in theory be a genetic marker for selective personality or temperament characteristics solely through genetic linkage; in such a case, the health differences observed between the ATD- and MC-mothers could be a consequence of personality characteristics associated with the ATD-genotype. For this interpretation to be true, some explanation would be required for the negative effect on mothers' health only, when both the mothers and the fathers are ATD-gene carriers. Decisive empirical test of this interpretation lies beyond the current study's design.

c) Mental stress. A third interpretation is that the considerable mental stress resulting from the ATD-matter has affected both the mental and physical health reported by the ATD-mothers. Previous findings for this sample would support this interpretation: The mother was generally the parent who received the first news of the child's ATD during the postpartum period, and a majority of the ATD-mothers (78 %) reported having negative emotional reactions upon receiving this information (3). More than a third of the mothers were judged (per retrospective reports) as having had "psychic crisis reactions" (14). In contrast, the fathers were generally much less involved with the child's ATD both practically and emotionally, and the fathers generally reported responding less negatively

Table 7

Reported changes in health status, compared with 5-10 years earlier, among mothers reporting "poor" or "very poor" health at present

Variable and group	Worse now n (%)	Better/ same now n (%)	Missing data (n)
Poor present physical health			
ATD-mothers (n = 13)	7 (58)	5 (42)	(1)
Control mothers (n = 8)	2 (25)	6 (75)	
Poor present mental health*			
ATD-mothers (n = 13)	6 (50)	6 (50)	(1)
Control mothers (n = 6)	1 (20)	4 (80)	(1)

* Scale points (often and almost always mental problems) in Table 1.

and less strongly than did the mothers; the fathers were also less often judged as having shown a crisis reaction (9 %).

Furthermore, if maternal stress due to the ATD-matter were responsible for the reportedly poorer maternal health, then a worsening of health should have taken place after the child's ATD was discovered. Some results suggest this was so: While the total ATD-group did not differ significantly from controls on changes in health since that time (Tables 1 & 3), more worsening had occurred among the subgroup of ATD-mothers (vs. MC-mothers) reporting currently poor health. As shown in Table 7, worsening of health was reported by 58 % of the ATD-mothers (vs. 25 % controls) with poor physical health during the past year, and by 50 % of the ATD-mothers (vs. 20 % controls) reporting poor mental health during the past year.

In total, the most plausible interpretation of the origin of the significantly worsened mental and physical health reported by the ATD-mothers for the year preceding follow-up seems to be that this was a consequence of the mental stress caused by the ATD-matter. Whether the mothers' awareness of their ATD-gene carrier status represented part of this stress is unknown; however, as both ATD-mothers and fathers are carriers, this awareness would have to have had especially negative importance to the mothers to account for the negative effects on maternal health only.

The negative effects of the ATD-matter on the ATD-mothers' reported health, plus the apparent lack of effect on the ATD-parents' experience of their current life situation, need to be included in the total cost-benefit analysis of the neonatal screening for ATD.

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